Re-initiation of Enzyme Replacement Therapy (ERT) During Pregnancy in a Patient with Gaucher Disease Type 1: A Case Report

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Abstract:

Introduction: Gaucher disease type 1 (GD1) is a rare, inherited lysosomal storage disorder caused by a deficiency of the enzyme glucocerebrosidase, leading to glucocerebroside accumulation in macrophages. This results in organomegaly, cytopenias, and bone abnormalities. Enzyme replacement therapy (ERT) is the standard treatment for GD1. Approximately 100 cases of GD1 treatment with ERT during pregnancy have been reported to date; however, there is a lack of documented cases concerning the re-initiation of ERT during pregnancy after a previous discontinuation.

Case Presentation: We present the case of a 37-year-old female patient with GD1, diagnosed in 2008 and treated with Cerezyme until 2017. The patient discontinued therapy due to the long distance to the enzymatic treatment center. In 2022, at 8 weeks of gestation, she presented to the Department of Internal Medicine with thrombocytopenia (approximately 50,000/μL) and frequent nosebleeds. After qualification and a 4-week waiting period for approval from the Ultra-Rare Diseases Team, Cerezyme treatment was re-initiated at 16 weeks of gestation. Initially, platelet counts remained low. Around 26 weeks, an increase to 70,000/μL was observed, and by 36 weeks, platelet count reached 98,000/μL, ensuring adequate hemostasis for an operative delivery. At 39 weeks of gestation, she delivered a healthy boy via Cesarean section. The postpartum period was uneventful.

Conclusion: This case highlights the feasibility of safely and effectively re-initiating ERT during pregnancy in GD1 patients, even after a prolonged treatment interruption, which is crucial for stabilizing hematological parameters prior to delivery.

Keywords:

Gaucher disease type 1, Enzyme replacement therapy, pregnancy.