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Title: Pregnancy with Gaucher's Disease- A rare metabolic disorder

OBJECTIVE

We aim to contribute to the limited literature on pregnancy outcomes in patients with Gaucher's disease and provide valuable insights for healthcare providers managing similar cases.

INTRODUCTION

Gaucher disease (GD) is an autosomal recessive lysosomal storage disorder caused by β-glucocerebrosidase deficiency, leading to glucocerebroside accumulation and a range of visceral, hematologic, and skeletal symptoms, with five subtypes varying in severity and age of onset.

CASE REPORT

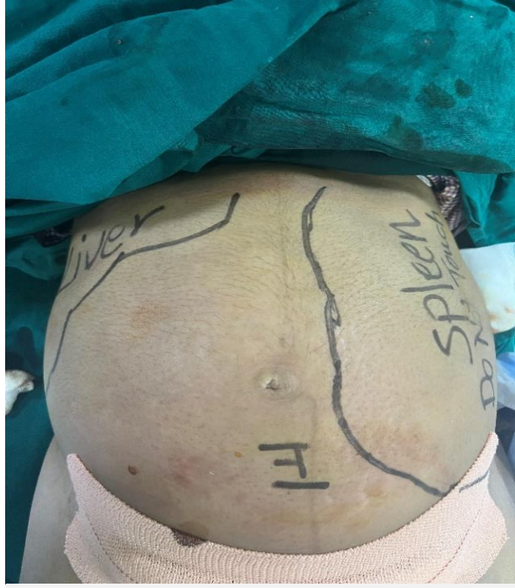
- A 30-year-old primigravida at 6.2 weeks gestation presented with hepatosplenomegaly and pancytopenia. Bone marrow biopsy confirmed Gaucher's disease.
- Enzyme analysis showed reduced glucocerebrosidase activity.
- Anaemia was treated with iron.
- Emergency LSCS was performed with no transfusion needed.
- Postpartum recovery was uneventful, and the baby was evaluated.

DISCUSSION

Type 1 Gaucher's disease, the most common non-neuronopathic form, may be diagnosed incidentally or with symptoms like painless splenomegaly, anaemia, and bleeding tendencies. Pathogenesis includes hepatomegaly, skeletal involvement, cytopenia, and bleeding risks due to Gaucher cell infiltration. Diagnosis is confirmed through glucocerebrosidase activity tests and PCR for GBA1 mutations. Treatment includes enzyme replacement therapy (ERT), which improves visceral and hematologic symptoms, and substrate reduction therapy (SRT) for selected patients. Pregnancy can exacerbate symptoms, and careful management is needed for anaesthesia due to hematologic and pulmonary complications.

CONCLUSION

In conclusion, this case highlights the importance of a thorough initial assessment and a multidisciplinary approach for managing pregnancy in Gaucher's disease, emphasizing the need for further research to better understand the condition during pregnancy.



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