

# A Case of Gaucher's Disease with Recurrent Fetal Loss and Antiphospholipid Antibody Syndrome

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## Introduction

Gaucher's disease is a leading cause for recurrent fetal loss in treated and untreated patients.<sup>1-3</sup> Gaucher's disease is associated with increased activity of immune system,<sup>4</sup> and raised auto-antibodies titers but there are a few reports in support of contribution of autoimmune phenomena in these patients.<sup>5</sup> In 1990, Haratz et al reported Coombs positive warm type autoimmune hemolytic anemia in a 23 year-old female with Gaucher's disease. They reported that further study on 72 patients with Gaucher's disease revealed another case of Coombs positive autoimmune hemolytic anemia in one of them and other autoimmune disorders in 17 patients of them.<sup>6</sup>

Sherer et al (2002) reported Coombs positive hemolytic anemia and antiphospholipid antibody syndrome in a 27 year-old female with Gaucher's disease who had successful outcome of pregnancy after treatment with combination of aspirin, low molecular weight heparin, prednisolone, and replacement of enzyme with Imiglucerase.<sup>7</sup>

## Case Report

I would like to present a case of Gaucher's disease with recurrent fetal loss and Antiphospholipid antibody syndrome.

A 31 year-old female was referred to our hematology clinic due to splenomegaly and pancytopenia. She had developed left upper quadrant pain and fatigue since 9 months ago with significant exacerbation during pregnancy which

ended to a living child. She had history of five pregnancies terminated with spontaneous abortions at 8-12 weeks. She had family history of anemia without taking any medical care in her father and sister. Physical examination showed pallor and splenomegaly (4cm below costal margin).

Laboratory findings revealed a white blood cell count of 3500/ $\mu$ L (Polymorphonuclear=65%, Lymphocyte=35%). Serological studies for HBS Ag, and HCV Ab were negative. Peripheral blood smear showed red blood cells with hypochromia, a few target cells, and a few ovalocytes. Bone marrow aspiration revealed hypocellular marrow, cells containing "crinkled paper" cytoplasm, and glycolipid-laden macrophages. Hence, diagnosis of Gaucher's disease was made. Immunohistochemistry (IHC) on bone marrow biopsy of this patient demonstrated positive CD68 in all large cells, as well as, positive lysozym in all large cells compatible with Gaucher's disease. The enzymatic assay revealed the level of glucocerebrosidase to be 0.5  $\mu$ mol/1h (with reference range >4  $\mu$ mol/1h). Our therapeutic plan was administration of Cerezyme (Imiglucerase).

Further assessment for other causes of recurrent fetal loss revealed antiphospholipid antibody syndrome with high titer of anti- $\beta$ 2-glycoprotein I antibody. Serologic tests for ANA, Anti-dsDNA, antiphospholipid antibody (IgM, IgG) and VDRL were negative.

## Discussion

Untreated and treated patients with Gaucher's disease are at risk for recurrent fetal loss. Diagnosis of Gaucher's disease should be considered in cases with unexplained and recurrent fetal loss. Hematologic and genetic study is recommended. Although recurrent fetal loss has been reported in patients with Gaucher's disease, we recommend assessment for antiphospholipid antibody syndrome in every patient with recurrent fetal loss and Gaucher's disease.

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