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Case Report

Gaucher's Disease Type 3 with Proteinuria: Case Report

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Abstract

Gaucher's disease type 3 with proteinuria is a rare case and had seen only one case in our hospital. This patient has severe anemia, leukopenia, thrombocytopenia, visceral organ enlargement and also has developmental retardation. He came with severe condition with weakness, oliguria and poor appetite. He also has yellowish brown pigmentation skin on extremities.

Even we didn't do bone marrow aspiration, genetic study or biopsy on this case but under appropriate discussion in team, we still think Gaucher's disease with proteinuria. We still learn more from this case for next diagnosis and management.

This case was provided supportive care with antibiotic treatment for sepsis, electrolyte correction, rehydration, steroid therapy, folic acid supplementation and social counseling.

Keywords: *Leukopenia; Thrombocytopenia; Enlargement; Retardation; Proteinuria*

Introduction

Gaucher's disease is a genetic disease in which a fatty substance (lipid) accumulates in cells and certain organs. It is caused by a hereditary deficiency of the enzyme glucocerebrosidase (acid beta glucosidase). Glucocerebroside can collect in the spleen, brain, lungs, kidneys and bone marrow. The National Gaucher Foundation state reported that 1 in 100 people in US population is a carrier: Gaucher's disease type 1: 1/40,000, Gaucher's disease type 2: no data and Gaucher's disease type 3: 1/50,000 in northern Swedish.

Case Report

A 10 years old boy had been sick 15 days with nasal discharge, low fever, sore throat, no vomiting and poor appetite. On the last 6 days the patient developed to poor urine output, both lower leg edemas, poor urine, abdominal pain, weakness and admitted for further management. He always presents in hospital with hematologic problem and hospitalized at AHC due to Hepatitis, Leukopenia, Anemia, and neutropenia. He is the second of three siblings; his elder and younger sisters are well.

On examination was found pale, lower legs edema, weakness and dry skin. There was hepatosplenomegaly with palpable liver 2 cm and spleen 1.5 cm below costal margin. He has hyporeflexia on both lower legs and slightly delay speech as mother's mention. He also has yellowish brown pigmentation skin on extremities.

Investigations revealed following: Hb 103, Hct 33%, RBC 4.36, MCV/MCH 75/24, Platelets 09, WBC 1.4, Neutro 0.4, Lympho 1.0, Malaria negative, reticu 0.9, Blood film: poki+, target cells +, burr cells+, acantocyte++. Electrolytes: Na 125, K 4.74, Cl 101, iCa 0.90, Ca 1.75, Urea

6.3, Creat 63, GGT 36, ALT 39, Total protein 60, Albumin 35, CRP 2.6. Urinalysis: Protein 100, WBC 15, no bacteria and other laboratory tests: ASO < 200, SLE negative, Blood culture negative in 48 h. Bone marrow aspiration and genetic study were not done with this case.

This case was provided supportive care with antibiotic treatment for sepsis, electrolyte correction, rehydration, steroid therapy and folic acid supplementation.

Discussion

Gaucher's disease is a rare case and had seen only one case in our hospital. Diagnosis was done with appropriated laboratory tests and discussion. This disease was treated with supportive care, supplementary vitamins and family explanation.

There are 3 subtypes of Gaucher's disease have been described and cases of different subtypes within one family have also been reported [1-6].

Type I disease has malignant childhood, benign adult, moderate-severity variable course variants.

Type II disease presents in infants and toddlers with clinical signs of severe opisthotonus, trismus, dysphagia, laryngeal spasm, general spasticity and developmental delay.

Our patient has severe anemia, leukopenia, thrombocytopenia, visceral organ enlargement and also has developmental retardation. He came with severe condition with weakness, oliguria and poor appetite. Even we didn't do bone marrow aspiration, genetic study or biopsy on this case but under appropriate discussion in team, we still think Gaucher's disease with proteinuria. We still learn more from this case for next diagnosis and management.

Conclusion

Gaucher's disease with proteinuria is a rare case and had seen only one case in our hospital. Diagnosis was done with appropriated laboratory tests and discussion. This disease was treated with supportive care, supplementary vitamins and family explanation.

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