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Splenic Artery Aneurysm: A Rare Complication of Type 1 Gaucher Disease

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Introduction
Gaucher disease (GD) is an autosomal recessive lysosomal disease due to acid betaglucosidase enzyme deficiency. The clinical manifestations of GD result from the accumulation of lipid-laden macrophages in the spleen, liver, bone marrow, bone, and other tissues.
GD may be complicated by conditions such as pulmonary hypertension, cirrhosis, spleen infarction, as well as hematological malignancies. Splenic artery aneurysms (SAA) account for more than half of all visceral aneurysms and are the third most common intra-abdominal aneurysm. It can be asymptomatic even if it reaches large sizes. In addition, the diagnosis of SAA may be difficult in GD because of the tortuous splenic artery due to splenomegaly. We presented a case of type 1 GD complicated by multiple myeloma and splenic artery aneurysm.

Case
A 56 years old male patient applied with a complaint of fatigue 20 years ago, splenomegaly and thrombocytopenia were detected, did not have a definite diagnosis at those years. Bone marrow biopsy was performed in 2017 and showed histiocytic cell infiltration which was compatible with Gaucher's disease. Also 82x72 mm splenic artery aneurysm was detected in the abdominal MRI performed at that time, and the aneurysm was repaired. GD was not given enough importance and specific treatment for GD could not be initiated.

The patient was referred to our clinic for GD four months ago. In enzyme measurement; plasma chiotriosidase was 586 µmol/L.hour, beta glucosidase (leukocyte) was 0.8 nmol/mg/hour. In genetic analysis, 1226A >G (rs76763715) (p.N409S) and 1193G>T (p.R398L) were found to be heterozygous mutation.

Dual energy X-Ray absorptiometry revealed osteopenia in the femoral neck. In volumetric MRI, the size of the spleen was 30x10x18 cm (2700 ml; 8.65 fold increase), and the liver was 21x15x20 cm (3150 ml; 1 time). In addition, urinary protein excretion was 1.5 g/day, serum immunoelectrophoresis for plasma cell dyscrasia revealed increased levels of kappa light chain (951 mg/L; N: 6.7-22.4 mg/L), lambda light chain (71.1 mg/L; 8.3-27 mg/L). Bone marrow biopsy was planned with suspicion of multiple myeloma.

Discussion
Although the prevalence of SAA in the general population is variable, such as 1-10.4% in autopsy series, the prevalence of SAA due to Gaucher's disease was rarely reported as 2% in a study. In its pathophysiology, increased intra-abdominal pressure caused by long-term splenomegaly, increase in flow turbulence due to portal hypertension, and loss of flexibility in the arterial wall structure caused by pathological macrophage infiltration can be counted. There is no difference in the treatment of SAA associated with GD. The risk of rupture can be reduced by reducing the flow

turbulence and pressure effects in the artery due to the reduction of spleen volume with enzyme replacement therapies. However, screening of SAA in GD is important in terms of early recognition of complications that may develop due to aneurysm rupture

Splenic artery aneurysm

