A Case of Systemic Mastocytosis with associated clonal hematological non-mast cell lineage disease

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Introduction: Advanced systemic mastocytosis can damage to multiple organs, such as liver, spleen, and gastrointestinal tract. In some of these patients, transformation to systemic mastocytosis with associated clonal hematological non-mast cell lineage disease(SM-AHNMD) is seen.

Case report: A 60-year-old woman underwent abdomen ultrasonography as a routine health examination. Unexpectedly multiple intra-abdominal lymphadenopathy was found. The computed tomography(CT) showed cystic duct wall thickening, multiple lymphadenopathy, collapsed gallbladder, and splenomegaly(Fig.1). She had chronic diarrhea, but not fever and abdominal pain, The initial laboratory test was white blood cell count(WBC) 9200/uL, Hemoglobin 11.5g/dL, platelet count 418000/uL. Endoscopic ultrasound(EUS)-guided celiac trunk lymph node biopsy(Fig.2) result was histiocytic and eosinophilic infiltration. Diagnostic laparoscopic cholecystectomy was done. Biopsy revealed lymphoesinophilic cholecystitis. In accrodance with eosinophilic cholecystitis, prednisolone 30mg was prescribed for 1 month. Lymph node slightly decreased at CT. After tappering prednisolone to 5mg, increasing size of lymph node, ascite, and hepatomegaly was found at CT. EUS-guided peri-pancreatic lymph node biopsy was done. There was no other finding than previous biopsy. Laparoscopic mesentery lymph node biopsy revealed primary follicle. Colonoscopy was done due to chronic diarrhea. There was diffuse yellowish mucosal lesion(Fig3). which revealed eosinophilic colitis. At that time, WBC increased to 14900/uL. Eosinophil count increased over 1000/uL, Platelet count was still high, 396000/uL. To differentiate systemic mastocytosis and histiocytosis, bone marrow biopsy was done. There was spindle-shaped mast cell(2.2%). CD117, CD2, CD25, vimentin immunostain was positive. KIT gene mutation was found. Patient was diagnosed to SM-AHNMD. She is followed up by CT without chemotherapy.

Conclusion: In case of prolonged multiple lymphadenopathy with organomegaly and abnormal complete blood cell count, not only lymphoma but also systemic mastocytosis and histiocytosis must be considered. And early bone marrow biopsy is recommended.

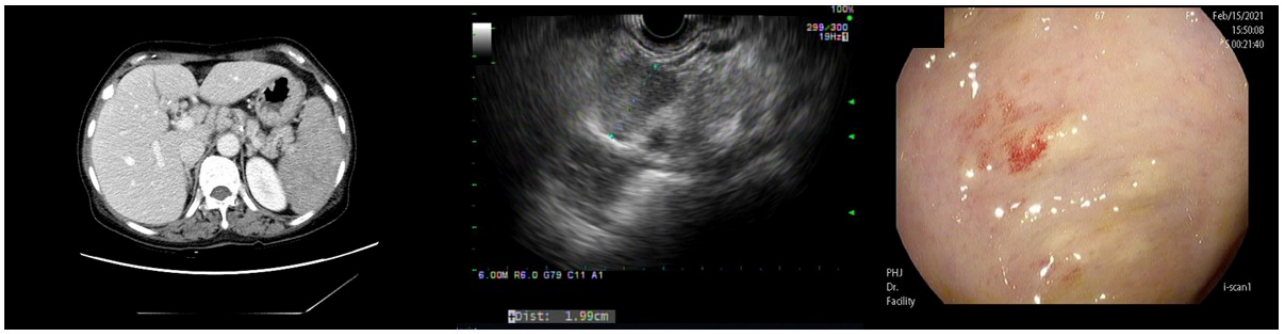

Figure1. Abdomen CT

Figure2. Endoscopic ultrasound

Figure3 Colonoscopy