Gastric cancer with very rare mutations in MSH2 and PMS2 successfully treated with nivolumab

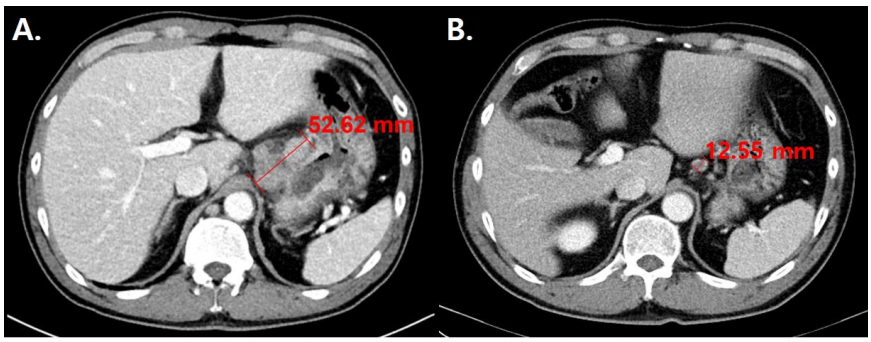
서울아산병원 내과[1], 서울아산병원 종양내과[2]

이보라[1], 류민희[2]

Background: We report a case of advanced gastric cancer with very rare mutations in MSH2 and PMS2, who has been successfully treated with nivolumab, an immune checkpoint inhibitor.

Case: A 49-year-old male presented with abdominal cramping pain and melena. A 6cm ulcero-infiltrative tumor mass in gastroesophageal junction was detected by esophagogastroduodenoscopy. The pathology of the tumor mass was poorly differentiated adenocarcinoma. Genetic testing by next generation sequencing showed MSH2(X426_splice), PMS2(K580*) mutations and microsatellite instability high (MSI-high) status. The computed tomography scans revealed multiple metastatses in abdominal lymph nodes and left supraclavicular lymph node. The patient received combination chemotherapy which consists of oxaliplatin 130mg/㎡ on day 1, capecitabine 1000mg/㎡ on days 1-14, and nivolumab 360mg on day 1, every 3 weeks. Due to peripheral neuropathy, oxaliplatin was discontinued at cycle 6 and then capecitabine and nivolumab were administered continuously. The patient achieved a partial response, and treatment has been ongoing until now for 2 years and 3 months.

Discussion: MSI-high patients are known to have a higher tumor mutational load and neoantigens, which make tumors more immunogenic. In those patients, immune checkpoint inhibitors are more effective. The current patient harboring very rare mutations in MSH2 and PMS2, which led to MSI-high status, shows a prolonged response to nivolumab-containing regimen.



A. The largest abdominal lymph node at the start of the chemotherapy
B. The evaluation of the disease after 2 years and 3 months