The Sequelae of Lynch Syndrome

Lynch syndrome is an autosomal dominant disorder and one of the most well-known hereditary cancer syndromes. Lynch syndrome significantly increases the risk of developing colorectal cancer, as well as an increased risk of developing other types of cancer, due to alterations in genes involved in DNA mismatch repair. The most common associated cancers are colorectal, endometrial, ovarian, bladder, stomach, and prostate. A unique case is presented here of a patient with Lynch syndrome that developed over 5 different malignancies over the course of his life and the sequelae of the malignancies, which lead ultimately to his death.

Patient is a 69- year-old male with past medical history significant for Lynch syndrome, prior ischemic stroke, and HFrEF requiring ICD placement who presented with complaints of abdominal pain that was worst near his urostomy site associated with nausea/vomiting, and diarrhea. Of note, throughout his lifetime, patient had gone through countless procedures and treatment for colon cancer s/p partial colectomy with reversed colostomy, renal cell carcinoma s/p left sided nephrectomy, bladder cancer s/p urostomy, papillary thyroid cancer s/p near total thyroidectomy, basal cell carcinoma, malignant neoplasm of lung, and newly diagnosed brain lesions concerning for metastatic disease. Vitals on arrival only showed a fever of 101.1F. Initial CT abdomen/pelvis was unremarkable. Urinalysis was concerning for complicated UTI and the patient was started on broad spectrum antibiotics given immunocompromised status and indwelling urostomy. Blood cultures grew Enterococcus faecalis in 2 of 2 bottles so Infectious Disease was consulted for urostomysourced bacteremia. Patient continued to deteriorate and developed sepsis, requiring multiple pressors and antibiotics broadened to micafungin and zosyn. Additional bacteremia sources, such as the Mediport and ICD, were recommended strongly to be removed but patient declined. Patient then developed pneumoperitoneum, ileus, renal failure, respiratory failure, and lower GI bleed throughout the hospital stay. Even knowing that the prognosis was extremely guarded, the patient was hopeful that he would make a recovery as he had beaten multiple cancers throughout his lifetime. After numerous conversations with palliative medicine and treatment teams, the patient and family decided on hospice. Unfortunately, the patient passed away prior to discharge to hospice.

The patient's Lynch syndrome increased the patient's lifetime risk of developing cancer and ultimately led to his death. In this patient, the suspected source of bacteremia, that led to the cascading events of the patient's death, was most likely the urostomy, required after cystectomy secondary to bladder cancer. The most interesting aspect of this case is the sheer number of malignancies developed over the patient's lifetime as well as the type of malignancies. Thyroid cancer is not usually considered to be associated with Lynch syndrome, with only a handful of known cases. This supports the importance of being vigilant and thinking of possible familial cancer syndromes if the history fits, even when the cancer is not typically associated with the various syndromes. The rare presence of thyroid cancer, along with the multiple other malignancies, in this Lynch syndrome patient truly makes this case unique.