

Promising Result with The Use of Immunotherapy to Treat Lynch Syndrome

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CASE REPORT

A 38-year-old male initially presented with complaints of abdominal pain with extensive history of changes in bowel habits. The patient's medical history included a prior motor vehicle accident and hypertension. After the patient presenting with complaining of Abdominal pain in ED A CT of abdomen was performed showing a necrotic appearing mass inseparable from the sigmoid colon and left hydroureteronephrosis that was treated with a 26cm double- J ureteral stent. He was also had a biopsy which revealed a poorly differentiated sigmoid colon adenocarcinoma, classified as at least stage IIIC after staging was complete/. Given the extent of the tumor, he was initially deemed unrespectable by surgical oncology. The tumor was MSI-unstable with loss of MSH2 protein. He was thus started on Dostarlimab 500mg given IV every 3 weeks. After 6 cycles of therapy, there was significant tumor response allowing for en bloc resection of his cancer. Final pathology showed a complete response to therapy and no viable tumor was noted. This was consistent with a pathological complete response (pCR). He has remained on Dostarlimab and will complete 1 year of therapy as recommended by our GI tumor board without significant complications.

Interestingly, the patient noted a family history for Lynch syndrome with his maternal aunt who is an identical twin of his mother. His mother was never tested for Lynch syndrome, but the patient did test positive for germline MSH2 mutation after meeting with genetics.

DISCUSSION

Lynch syndrome, a hereditary cancer syndrome characterized by germ line mutations in DNA mismatch repair genes such as MSH2 as noted in this case. It, presents intricate challenges in both diagnostic evaluation and therapeutic management. The diagnosis of Lynch syndrome in this patient was of particular significance, as it impacted the therapeutic aspects of his care. This predicted a high probability of response with immunotherapy as front-line treatment as compared to traditional chemotherapy. In addition, it necessitates increased cancer screening and surveillance due to the syndrome's association with an elevated risk of multiple cancers. Challenges arose in choosing the optimal duration of treatment as there is a lack of clinical trial data with resected colorectal cancer and the use of immunotherapy in the adjuvant setting especially in patients who achieve a pCR with neoadjuvant therapy.

CONCLUSION

This case report illustrates the complexities related to the diagnosis and management of a 38-year-old male presenting with initially unresectable poorly differentiated sigmoid colon adenocarcinoma and Lynch syndrome. Multidisciplinary collaboration and a tailored approach are crucial for optimizing outcomes and providing personalized care. Early genetic testing for MSI status and Lynch syndrome is imperative in patients who have colorectal cancer and should be the standard of care. This case highlights the significance of personalized care in the management of patients diagnosed with Lynch syndrome and its associated malignancies and promising outcome with the use of immunotherapy and the importance of collaboration among health care team in delivery best care for the patient survival.