Journal of Patient Experience
Volume 10: 1-4

DOI: 10.1177/23743735231189348
journals.sagepub.com/home/jpx
Sage

"Lynch Syndrome"—From Healer to a Vulnerable Patient: A Transformative Odyssey

Aisha Khalid, MD[1]

Abstract

Lynch syndrome, an autosomal dominant genetic disorder, increases the risk of certain cancers, notably colorectal cancer. Early genetic testing and surveillance can significantly reduce associated morbidity and mortality. However, young patients face significant challenges navigating the healthcare system, with the psychological impact often neglected. Coping with the hereditary condition and elevated cancer risk can lead to distress, anxiety, and depression. Healthcare providers tend to focus on physical aspects, disregarding psychological well-being, leading to reduced treatment compliance and satisfaction. This case highlights the need for multidisciplinary teams and mental health support, emphasizing patient-centered care and support systems within the healthcare system.

Introduction

Lynch syndrome is caused by a mutation in one of the DNA mismatch repair genes, most commonly MLH1, MSH2, MSH6, or PMS2.[1] Individuals with Lynch syndrome have a high lifetime risk of developing colorectal cancer and other cancers, such as endometrial, ovarian, gastric, pancreatic, urinary tract, and brain tumors.[2] Early identification through genetic testing and surveillance is crucial for improved outcomes. Despite the increasing awareness of Lynch syndrome and its genetic testing, the challenges faced by young patients in navigating the healthcare system from diagnosis to management remain significant. The psychological impact of Lynch syndrome on young patients is often overlooked or inadequately addressed within the healthcare system. Coping with the knowledge of a hereditary condition and the increased risk of cancer can cause significant distress, anxiety, and depression in affected individuals. Unfortunately, many healthcare providers may focus primarily on the physical aspects of Lynch syndrome management, neglecting the psychological well-being of their patients. This lack of psychological support further exacerbates the emotional burden experienced by young patients, potentially leading to reduced treatment compliance, decreased overall satisfaction with the healthcare system, and poorer long-term outcomes.

This case reflects the challenging journey of a young physician diagnosed with Colon cancer (Lynch syndrome) in the prime of her career. It has highlighted the importance of mental health support by recognizing flaws in the healthcare system and emphasizing the significance of patient-centered care and support systems.

My Personal Experience

Background

Despite having achieved significant success in my career as a medical trainee, my relentless pursuit of knowledge led me to embark on a new adventure: pursuing a master's degree in research at the esteemed Harvard Medical School in the United States. This life-changing decision necessitated leaving behind everything I knew in Australia and embracing the challenge of immersing myself in a new culture. It was a mixture of trepidation and exhilaration as I embarked on this daunting yet invigorating journey. However, amidst the excitement, the stress associated with relocating took a toll on my physical and mental well-being.

Diagnosis

I started experiencing intermittent pain in my right iliac fossa for a couple of weeks. The pain became so persistent that I sought medical attention during a holiday back home. While waiting in the emergency room for the results of a computed tomographic (CT) scan, a surgical registrar, with a grave

[1] Harvard Medical School, Boston, MA, USA

Corresponding Author:
Aisha Khalid, Harvard Medical School, 25 Shattuck Street, Boston, MA 02125, USA.
Email: aisha.khalid000@gmail.com

expression, delivered unexpected news: I had been diagnosed with likely colon cancer. The news came as a shock, particularly because I had no family history of the disease, was still in my early thirties, and had never anticipated facing such a severe illness at this stage. I have always taken great care of my health. Initially, there was a possibility that the lesion observed in my scans could be lymphoma. However, after a battery of tests, including staging CT scans, blood tests, tumor markers, and a colonoscopy, it was confirmed that I was dealing with obstructing colon cancer. Although there was a slight delay in scheduling the colonoscopy, a few calls to my superiors helped expedite the process, and I was promptly scheduled for surgery after the holidays.

Preoperative Management

With the long weekend fast approaching, I was allowed to return home for preoperative management following the Enhanced Recovery After Surgery protocol. However, to my disappointment, I discovered that the preoperative drinks provided were expired. I promptly informed the Colorectal nurse about the issue, and as a result, I was advised to refrain from consuming the drinks altogether. While this situation initially caused me some concern, I attributed it to a minor human error and decided to let it go.

Postoperative Management

My postoperative journey began without patient-controlled analgesia (PCA) and only oral pain medication, specifically Endone. This unexpected change threw me off guard as I woke up after surgery experiencing intense pain. It took the nursing staff approximately 2 hours to arrange the PCA, which was later apologized for by an anesthetist.

Throughout the day, my interactions with the surgical fellow and junior doctors were limited to a brief 5-minute morning ward round. While I understood their busy schedules, I received no updates regarding my blood test results or other crucial information. Concerned about my well-being, I self-diagnosed refeeding syndrome when I experienced extreme lethargy despite diligently following all postoperative recovery instructions. This lack of attention and communication from the medical team became another source of disappointment during my recovery.

One of the most uplifting aspects of my postoperative recovery was the exceptional empathy displayed by the nursing staff. Their compassionate care and understanding provided comfort during a challenging time. During the consultant ward round, the operating surgeon shared his own battle with malignancy. This genuine and relatable conversation created an instant connection and comfort, further enhancing my recovery experience.

Despite being in the midst of my hospital stay, I faced the pressure of a looming deadline for my capstone project at Harvard. Determined to fulfill my commitments, I continued working on it while in the hospital. Eventually, I was discharged home while awaiting the histopathology results, which carried the possibility of a Lynch syndrome diagnosis. I had already arranged an appointment with the oncologist to ensure proper follow-up and care.

Chemotherapy Vs No Chemotherapy?

During my follow-up appointment with the oncologist, I was informed of the likelihood of Lynch syndrome (pending genetic testing). The TNM staging of my colon cancer was determined to be T3N0M0, and the tumor was classified as poorly differentiated adenocarcinoma. However, given my background and specialization in gastroenterology, specifically in Lynch syndrome, I found it confusing that chemotherapy was recommended as the primary treatment option. This contradiction prompted me to seek a second opinion. The second opinion was the exact opposite of the first one. Getting a genetics referral was lengthy, taking nearly 2 months to secure the first appointment. Subsequently, arranging the necessary blood testing and obtaining the genetic results took 3 additional months.

Fertility Preservation

Considering my situation, my initial oncologist advised chemotherapy as the primary treatment option. In addition, due to the unavailability of a public fertility specialist for the next 6 months, a referral was made to a private fertility specialist to discuss fertility preservation options, given my age. Despite being uncertain about the management plan, I found myself in a situation where I had to pay a significant amount out of pocket for the fertility preservation process. The fear of losing the battle against the biological clock pushed me to take this financial step, even without complete clarity on the treatment path.

Psychological Support

As a young aspiring gastroenterologist, being diagnosed with Lynch syndrome while having a nonmedical partner and no other family support was an overwhelming experience. Within 3 months, everything seemed to be happening so quickly. However, what struck me the most was the lack of attachment to any support group or psychological assistance throughout this challenging period by my hospital or any of my treating physicians because the exact diagnosis took a long time. It was truly disheartening. I didn't realize the extent of my need for help until I noticed a significant change in my emotional state. Suddenly, I became excessively irritable, frequently crying and arguing over trivial matters, which was entirely out of character. At that point, I realized the importance of seeking assistance and support.

Long-Term Management

Currently, I am still awaiting connection to the Lynch syndrome program in Australia for both my family's genetic testing and my own long-term management plan.

Discussion

Lack of Multidisciplinary Team for the Management Plan

Multidisciplinary team (MDT) meetings are crucial for managing colon cancer with Lynch syndrome due to the complex nature of the disease and the need for comprehensive, individualized care. MDTs bring together experts from various specialties, including surgeons, medical oncologists, radiation oncologists, pathologists, genetic counselors, and other healthcare professionals. Through collaborative decision-making and collective expertise, MDTs ensure accurate diagnosis, staging, and treatment planning tailored to the specific needs and challenges of Lynch syndrome. They facilitate consensus building, provide opportunities for second opinions, and promote ongoing monitoring and adjustments to the management plan.[3]

This aspect was deficient in the patient's management, which prompted the author to seek a second opinion. If the treating physician had conducted a MDT meeting, it could have potentially led to a more comprehensive and well-coordinated management plan.

Delays in Genetic Testing and Appointments

Delays in acquiring genetic appointments and testing for patients with possible Lynch syndrome can significantly affect long-term management plans. Timely genetic testing is essential to confirm the diagnosis and identify specific Lynch syndrome mutations, which are crucial in risk assessment and management strategies. Delayed genetic testing can result in prolonged periods without definitive information, leading to delayed implementation of appropriate surveillance measures and interventions. This delay may increase the risk of undetected cancers and hinder the initiation of preventive measures in at-risk individuals and their families. Additionally, delayed genetic appointments and testing can impact the timely provision of genetic counseling and family screening, further impeding the comprehensive management of Lynch syndrome. Efforts should be made to minimize delays, optimize access to genetic services, and ensure efficient coordination among healthcare providers to support timely and appropriate management for individuals with Lynch syndrome.[4,5]

Lack of Public Appointments for Fertility Preservation for Cancer Patients

The lack of available public appointments for fertility preservation in cancer patients, particularly those with Lynch syndrome, can have significant financial and mental health effects. Fertility preservation procedures can be costly, and the absence of publicly funded appointments may place a financial burden on individuals seeking these services. The out-of-pocket expenses associated with private appointments and procedures can be substantial, adding to the financial stress experienced by cancer patients. Furthermore, the emotional toll of facing a cancer diagnosis and the potential loss of fertility can be overwhelming. The additional challenges of navigating limited appointment options and financial considerations can exacerbate these individuals' anxiety, stress, and emotional distress. This case highlights the importance of addressing these barriers and providing accessible and affordable fertility preservation options to support the holistic well-being of cancer patients, including those with Lynch syndrome.[6]

Lack of Psychological Support

Psychological support, including counseling, education, and psychosocial interventions, can help patients cope with the emotional challenges associated with Lynch syndrome, enhance their psychological well-being, and improve their overall quality of life. It also facilitates informed decision-making regarding risk management strategies, genetic testing, and cancer screening protocols. The National Comprehensive Cancer Network (NCCN) guidelines emphasize the importance of psychological support and recommend that individuals with Lynch syndrome receive appropriate counseling and psychosocial care.[7] The patient felt lacking this in her journey and established the dire need for it.

Poor Communication Skills

Poor communication skills in cancer patient management can have detrimental effects on both the medical and mental health aspects of patient care. Inadequate communication can lead to a lack of shared understanding, compromised patient–provider relationships, and suboptimal decision-making processes. Studies have shown that poor communication between healthcare providers and cancer patients is associated with increased patient anxiety, decreased patient satisfaction, and lower adherence to treatment plans.[8] It can also contribute to higher levels of psychological distress, emotional burden, and decreased quality of life for patients.[9] Conversely, effective communication has been linked to improved patient outcomes, increased patient satisfaction, and better emotional well-being.[10] The physician shared her experience about how poor communication skills had affected the timely pain management and the missed diagnosis of refeeding syndrome, leading to mental distress for the patient.

Conclusion

Lynch syndrome in young patients poses unique challenges, including difficulties in diagnosis, complex management, and inadequate psychological support. A physician's personal experience with Lynch syndrome and colon cancer highlights the need for personalized care, effective communication, and comprehensive support systems. A multidisciplinary approach

is crucial to address patients' physical, emotional, and psychological needs. By applying these lessons, we can strive to enhance patient care and outcomes for those with Lynch syndrome and similar genetic disorders.

Declarations of Conflicting Interests

The author declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Ethical Approval

This is a narrative review, so Ethics approval is not warranted.

Funding

The author received no financial support for the research, authorship, and/or publication of this article.

ORCID iD


Aisha Khalid https://orcid.org/0000-0001-8711-7962

References

1. Edwards P, Monahan KJ. Diagnosis and management of Lynch syndrome. *Frontline Gastroenterol*. 2022;13(e1):e80-7.
2. Møller P, Seppälä T, Dowty JG, et al. Colorectal cancer incidences in Lynch syndrome: a comparison of results from the prospective lynch syndrome database and the international mismatch repair consortium. *Hered Cancer Clin Pract*. 2022;20(1):36.
3. Vasen HFA, Blanco I, Aktan-Collan K, et al. Revised guidelines for the clinical management of Lynch syndrome (HNPCC): recommendations by a group of European experts. *Gut*. 2013;62(6):812-23.
4. Balmaña J, Balaguer F, Cervantes A, Arnold D. ESMO Guidelines working group. Familial risk-colorectal cancer: ESMO clinical practice guidelines. *Ann Oncol*. 2013;24(Suppl 6):vi73-80.
5. Stoffel EM, Mangu PB, Gruber SB, et al. Hereditary colorectal cancer syndromes: American society of clinical oncology clinical practice guideline endorsement of the familial risk-colorectal cancer: European society for medical oncology clinical practice guidelines. *J Clin Oncol*. 2015;33(2):209-17.
6. Loren AW, Mangu PB, Beck LN, et al. Fertility preservation for patients with cancer: American society of clinical oncology clinical practice guideline update. *J Clin Oncol*. 2013;31(19):2500-10.
7. Weiss JM, Gupta S, Burke CA, et al. NCCN Guidelines® insights: Genetic/familial high-risk assessment: colorectal, version 1.2021. *J Natl Compr Canc Netw*. 2021;19(10):1122-32.
8. How does communication heal? Pathways linking clinician–patient communication to health outcomes. *Patient Educ Couns*. 2009;74(3):295-301.
9. Faller H, Schuler M, Richard M, Heckl U, Weis J, Küffner R. Effects of psycho-oncologic interventions on emotional distress and quality of life in adult patients with cancer: Systematic review and meta-analysis. *J Clin Oncol*. 2013;31(6):782-93.
10. Jenkins V, Fallowfield L, Solis-Trapala I, Langridge C, Farewell V. Discussing randomised clinical trials of cancer therapy: Evaluation of a cancer research UK training programme. *Br Med J*. 2005;330(7488):400.