I Have Lynch Syndrome

Dear Chief Editor

Acta Medica International
Four years ago I was diagnosed with Lynch syndrome; an autosomal dominant inherited disorder of cancer susceptibility caused by germline mutations in the DNA mismatch repair genes. This mutation has substantially increased risks of early onset cancers of the colon and rectum, stomach, intestine, liver, pancreas, gallbladder ducts, upper urinary tract, brain, skin, and if you are male, the prostate. Women with Lynch syndrome are also at risk of developing cancer of the endometrium, ovaries and breasts.

There are an estimated 1,000,000 Lynch carriers in the United States alone, yet only 5% have been diagnosed. Additionally, it is believed that approximately 5% of all colon cancers in the United States can be attributed to Lynch syndrome.

There is a long history of colon cancer in my family and I frequently brought this up with my doctors but any suggestion of Lynch syndrome and its implications was never considered. Lynch syndrome is not nearly as recognized as it must be given similar family histories, therefore it is a major part of my advocacy that Lynch syndrome be considered whenever a family history includes, in particular, early onset colon cancer.

My father had colon cancer in his forties but fortunately it was caught early and he managed to live into his seventies. Various relatives on my paternal side had colon cancer as well but also survived. Then my oldest brother died at the age of 36 from colon cancer, diagnosed too late to treat. I started to suspect a genetic component to my family's colon cancer and began having regularly scheduled colonoscopies while in my twenties.

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A few years ago my other brother developed colon cancer in his late forties. Fortunately his doctor knew about Lynch syndrome and recommended he be genetically tested. He tested positive and that is when I started to learn about Lynch. An early search brought me to a genetic counselor and I realized they are essential specialists in the genetic testing process. They are exceptionally aware that patients considering a genetic test are in a most vulnerable position, they are able to help direct patients through the process and demonstrate how to reach out to other family members to recommend they be tested as well. But specifically, they are extremely knowledgeable about the recommended annual Lynch screening protocols which doctors must follow in order to prevent or detect cancer as early as possible.

I, too, tested positive and almost immediately underwent the recommended radical hysterectomy and bilateral salpingo-oophorectomy to reduce the chances of malignancy to my reproductive organs. This uncovered another knowledge gap in my Lynch journey. Being thrust, overnight, into early menopause is fraught with difficulties that my gynecologist neglected to mention - I went in short order from feeling a healthy and energetic 40 year old to sick, nauseated, lethargic, defeminized, and depressed. Ironically, I underwent the surgery to prevent malignancy but became physically ill from the loss of my ovaries. It took a year or more to tweak my hormone levels and I required a cocktail of other hormones as estrogen replacement was not enough - progesterone and testosterone were also prescribed after repeated visits to my gynecologist, and several other specialists whom I discovered through my own research.

Many women with Lynch syndrome undergo prophylactic surgery in order to prevent cancer in their reproductive organs. Various factors may impact a patient's quality of life following these types of surgeries and may hold a number of other health implications for other body parts and systems. These implications vary, and doctors are often only focused on saving a life or removing a cancer without giving much thought to how the quality of one's life may be impacted. Doctors need to realize that the "one size fits all" approach does not indeed fit all and should treat each patient in a unique manner - which may be their professional intention but does not always happen as much as it ought to. Gynecologists and gynecological oncologists, in particular, cannot overestimate hormone replacement for women who have had oophorectomies prior to menopause.

My hellacious experience with life *sans* ovaries along with the emotional roller coaster of a positive Lynch test result was the impetus for my blog, ihavelynchsydrome.com. In searching the web for information and support I discovered there was nothing out there and began to realize there must be others in a similar circumstance. My initial intention was to blog about the emotional aspects of having Lynch syndrome, to discuss my negative experience with having prophylactic surgery, try to offer support to other Lynch carriers whilst grappling with the terms of my mortality. And, worst of all, not knowing if my young son could be a carrier.

My own recovery would become a litmus test for many of my relationships. I felt anger and frustration because my extended family would not take the Lynch information from my genetic counselor seriously - they ignored my pleas to get tested, and preferred to simply not know what their genetic fates may hold. There were days I wanted to cease to exist, or at the very least, peel off my skin because I was physically and emotionally exhausted. Psychological and psychiatric intervention was needed. I required medication for anxiety and depression. It would take a few years for me to come to terms with my diagnosis, my mortality, my new body, and eventually occupy a much better, positive space in my life. I have found tremendous solace in Buddhism, acupuncture, meditation, my husband's understanding, my son's kisses, a few close friends, my "therapy" dog and with the help of the passage of time.

Rather than fretting over having Lynch syndrome, I have tried to assume some control of my genetic destiny as I strongly believe epigenetic factors are highly influential in regards to one's gene expression. I have become exceptionally vigilant with my health, eating only a plant-based diet and exercising frequently. Putting energy into advocacy work and helping others in turn helps me. These behavioral changes and insightful perspectives are where I seek comfort. Reading about other horrific genetic mutations, especially those which afflict children has given me tremendous perspective. I also think about those in the world who are truly living in misery and are suffering every single day simply because their basic needs are not met. Expressing gratitude for the many other positives in my life keeps me grounded.

Globally, there are too many variations regarding the frequency of screening for Lynch syndrome related cancers, which is yet another gap in the Lynch saga. In the United States an annual colonoscopy is highly recommended for those with Lynch syndrome but screening should not end there - my annual exam at the Mayo Clinic in Rochester, MN, includes the following:

- Blood test
- Urine test
- CA-125 blood test (as I no longer have my ovaries)
- Mammogram
- Colonoscopy with blue dye to detect flat polyps which are common in Lynch carriers
- Skin exam
- Endoscopy
- Pelvic ultrasound

Other issues that make tested for Lynch syndrome so difficult include patients who are scared of having a colonoscopy for several reasons: patients are unaware that colonoscopies can be done with sedation; most people complain about the taste and quantity of prep which they have to take; and people, especially men, are reluctant to undergo colonoscopies mostly due to the placement of the scope. Doctors need to take the time and make a concerted effort to discuss the details of what a colonoscopy entails with their patients.

Furthermore, cultural and ethnic differences need to be considered and addressed; genetic counselors and doctors need to specifically tailor discussion and treatment to accommodate different groups. Specific cancers may hold huge stigmas amongst certain groups, which in turn hold implications for whether or not a particular group will be screened. One example lies within the ultra-Orthodox Jewish community; many fathers are reluctant to have their daughters undergo genetic testing prior to marriage and prefer their daughters wait until after marriage. Among African-Americans it is a common misconception that colon cancer is the result of homosexual relations. Due to this social stigma, African-Americans are less likely to be screened and are more likely to die from colon cancer.

When doctors, specifically internists, gastroenterologists, and gynecologists, are taking a patient's family medical history and discover the following: three or more relatives have had colorectal cancer, cancer of the endometrium, small intestine, ureter or renal pelvis; two or more successive generations have been affected; and one or more relatives have been diagnosed before the age of 50, then this should give them tremendous pause. Doctors should refer these patients to a genetic counselor for testing. Genetic counseling, genetic testing, and vigilant annual screening can save lives.

My work has expanded quickly over the past few years and I am now applying to incorporate as a not-for-profit organization. My intentions are to continue to help those with Lynch syndrome by giving them hope and trying to decrease their despair. My greatest goal is to increase Lynch syndrome awareness amongst the global medical community - especially now since colon cancer is becoming more prevalent worldwide in people under the age of 50.

Lynch syndrome is not deemed a rare syndrome, merely underdiagnosed. Genetic counseling with genetic testing is extremely helpful for patients; genetic counselors can help a doctor improve a Lynch patient's chances of survival because they are fully aware of the palette of medical testing and may be able to provide a plethora of resources for the other issues including recommending looking for therapists and/or psychologists specializing in patients with cancer. Prophylactic surgery may prevent cancer but there are implications for removing body parts and quality of life issues may arise. The gravity of a Lynch syndrome diagnosis can be overwhelming for many and may hold implications for many aspects of a patient's life. The global discrepancies regarding Lynch syndrome screening need to be addressed and reevaluated.

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Editor's comments: Georgia Hurst is a patient advocate for those with Lynch syndrome and other hereditary cancer syndromes. Her strong family history of early onset colon cancer prompted her to seek out genetic counseling. She was tested positive for Lynch syndrome and began to assume some control over her genetic destiny by undergoing the prophylactic surgeries to prevent malignancy to her reproductive organs. The deep, negative psychological and physical impacts of removing her reproductive organs were the impetus for the blog: ihavelynchsyndrome.com. She frequently writes about the emotional implications of having Lynch syndrome. She recently received non-for-profit status for ihavelynchsyndrome,Inc.; The mission of publishing this letter to editor is to raise global awareness for hereditary cancer syndromes.

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