Dear Hereditary Cancer Listserv Members:

This month we are presenting a case study highlighting the variability that can be seen in families with Lynch syndrome.

**Case Study:** S.C. is a 46 y/o female with no personal history of cancer.

**Family history:**
- Sister: endometrial cancer at age 46 (deceased)
- Mother: endometrial cancer at age 61 (deceased)
- Maternal uncle: colon cancer at age 56
- No relatives tested previously for Lynch syndrome

**Current surveillance:** Annual mammograms; annual gynecological exams; since she is under age 50, no colonoscopy to date.

**Hereditary cancer testing:**
- This family includes multiple red flags for Lynch syndrome: the patient has two first-degree relatives with endometrial cancer, one under the age of 50, and a second-degree relative with another Lynch syndrome-associated cancer (maternal uncle with colon cancer).1
- Hereditary cancer testing (germline testing) for the MLH1, MSH2 and MSH6 genes revealed a deleterious mutation in MSH2, confirming Lynch syndrome in S.C.

**Impact on management:**
- S.C. was advised about her cancer risks and the recommended medical management for patients with Lynch syndrome. See [https://www.myriadpro.com/test-offerings/genetic-testing/colaris](https://www.myriadpro.com/test-offerings/genetic-testing/colaris). Ultimately, she elected to have a prophylactic hysterectomy and bilateral salpingo-oophorectomy and to begin annual colonoscopies.
- S.C. notified her two children and other relatives of the result. These relatives can undergo Single Site MSH2 testing, which will assess for the specific mutation identified in S.C.
- Not only did this test impact S.C.’s management, it will also impact the management and health of her family.

**Discussion:**

**Is this an unusual presentation for a Lynch syndrome family?** Although there were no cases of early onset (< 50 y) colon cancer, women with Lynch syndrome are as likely to present initially with a gynecologic cancer as with a colorectal cancer .2
References:

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