Case Study of PMS2 Lynch syndrome diagnosed by an OB-Gyn

This month we describe an actual case of Lynch syndrome identified by an OB-Gyn provider, based on an abnormal family history of cancer. The case highlights modified cancer risks and management recommendations for one of the more recently discovered Lynch syndrome genes, PMS2. The diagnosis has a potential impact on the management decisions for a patient presenting to her OB-Gyn with the common indication of contraceptive management.

Patient: 34 year-old female, annual visit/IUD placement
Personal history: no cancer or pre-cancerous polyps
Family history: Sister– colon cancer, 49 y; maternal grandmother– stomach cancer, 62

The patient’s OB-Gyn identified this family history as an abnormal screen and wanted to rule out Lynch syndrome. (Red Flags for Lynch Syndrome)

The OB-Gyn explained the red flags and implications to the patient and ordered Lynch syndrome testing (COLARIS®) after informed consent was obtained.

Result: Patient is positive for a deleterious mutation in PMS2, confirming Lynch syndrome with risks of up to 20% for colorectal cancer and up to 15% for endometrial cancer by age 70y. (Cancer risks for PMS2-associated Lynch syndrome are somewhat lower than those associated with other Lynch-related genes.)

Because the patient has Lynch syndrome, she should be managed at the hereditary risk level. NCCN guidelines suggest the following:

Colon: Colonoscopy every 1-2 years, starting at age 25-30. At 34, this patient is overdue for her first colonoscopy. Had she been managed based on family history alone, colonoscopies would have begun at 39 (10 y prior to sister’s age at diagnosis) and been repeated every 3-5 years.

Gynecological: Offer risk-reducing hysterectomy with salpingo-oophorectomy when childbearing is complete. Since the OB-Gyn just placed an IUD, he will need to discuss whether this patient’s family is complete or if the IUD is a temporary measure. Patients who choose to delay or decline surgery may consider endometrial and ovarian cancer screening, although there is no clear evidence to support this.

Other considerations: Upper endoscopy every 2-3 years, starting at age 30-35; annual urinalysis starting at age 25-30.

Beyond the patient: This patient’s family members can benefit from single site testing for the identified PMS2 mutation. Parents, siblings and children have a 50% chance of having the same mutation. Testing of minors is not generally recommended; rather, testing should be considered when medical management would change – in this case, as early as 25 y. Now that this patient has been identified as having Lynch syndrome, she and her family members can take proactive steps to help prevent additional cancers.
REFERENCES: