

Clinician Management Resource for *PMS2* (Lynch syndrome)

This overview of clinical management guidelines is based on this patient's positive test result for a *PMS2* gene mutation. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)¹ in the U.S. Please consult the referenced guideline for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decisions but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider, and may change over time.

| SCREENING/SURGICAL CONSIDERATIONS ¹ | AGE TO START | FREQUENCY |
|--|--|------------------------|
| Colorectal Cancer | | |
| Colonoscopy | 20-25 years old (or 2-5 years prior to the earliest colorectal cancer in the family, if it is diagnosed before 25 years) | Every 1-2 years |
| Endometrial and Ovarian Cancer | | |
| Endometrial: encourage prompt response to symptoms (e.g. abnormal uterine bleeding, postmenopausal bleeding). | Individualized | Individualized |
| Ovarian: educate women on the symptoms associated with ovarian cancer (e.g. pelvic/abdominal pain, bloating, difficulty eating, increased abdominal girth, etc.). | Individualized | Individualized |
| Consider option of prophylactic hysterectomy and bilateral salpingo-oophorectomy (BSO) for women who have completed childbearing | Individualized | N/A |
| Consider screening via endometrial biopsy | Clinician's discretion | Every 1-2 years |
| Transvaginal ultrasound and serum CA-125 may be considered. Data do not support routine ovarian screening. | Clinician's discretion | Clinician's discretion |
| Consider risk reduction agents | Individualized | Individualized |
| Gastric and Small Bowel Cancer* | | |
| Selected individuals/families or those of Asian descent may consider upper endoscopy with visualization of the duodenum. No clear evidence to support screening for gastric, duodenal, or small bowel cancer. Consider testing and treating <i>H. pylori</i> . | Beginning at age 40 | Every 3-5 years |
| Central Nervous System Cancer* | | |
| Consider physical/neurological examination | 25-30 years old | Every 12 months |
| Urothelial Cancer* | | |
| Selected individuals such as with a family history of urothelial cancer may consider urinalysis. There is insufficient evidence to recommend a particular surveillance strategy. | 30-35 years old | Every 12 months |
| Breast Cancer | | |
| Not enough evidence to support increased screening above average-risk screening recommendations or based on personal and/or family history. | Clinician's discretion | Clinician's discretion |

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|---|------------------------|------------------------|
| Prostate Cancer | | |
| Insufficient evidence to recommend earlier or more frequent screening | Clinician's discretion | Clinician's discretion |
| Other ¹ | | |
| Counsel for risk of autosomal recessive condition in offspring If both parents have a <i>PMS2</i> mutation, each of their children have a 25% chance to have constitutional MMR deficiency (CMMRD) syndrome. | Individualized | N/A |

* For *PMS2*, the risk of other Lynch syndrome-related cancers is reportedly low.

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines[®]) for Genetic/Familial High-Risk Assessment: Colorectal. V3.2019. © National Comprehensive Cancer Network, Inc. 2019. All rights reserved. Accessed December 26, 2019. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.