Memorial Sloan Kettering Cancer Center

患者及照护者教育

关于 PMS2 基因突变

本信息介绍了 *PMS2* 基因突变可能会对您和您家人产生的影响。

在本资源中，“家庭”一词指与您有血缘关系的人。 他们与您不属于婚姻或收养关系。 这类家庭成员也称为血缘亲属。

PMS2 基因通常有助于预防癌症。 该基因的突变会导致其停止正常运作。

重要的是要明白，这种基因发生突变并不意味着一定会罹患癌症。 这意味着您罹患某些类型癌症的风险会增加。 即使在同一个家族中，基因突变者的癌症类型也会有所不同。

如果出现 *PMS2* 基因突变，罹患癌症的风险有多大?

如果您有 PMS2 基因突变，这意味着您会患有一种称为林奇综合征的疾病。

Lynch syndrome increases your risk for certain types of cancers, including:

- Colorectal (colon and rectal) cancer
- Uterine (endometrial) cancer

Lynch syndrome may also increase your risk for other cancers, but this is less common. Examples of less common cancers linked to Lynch syndrome are:

- Ovarian cancer
- Stomach cancer
- Small intestine (small bowel) cancer
- Urinary tract cancer
- Pancreatic cancer
- Hepatobiliary tract cancer (cancer in the cells of the liver, bile ducts, and

gallbladder)

- Brain cancer
- Sebaceous carcinoma (cancer in the glands in your skin that make oil)

As we learn more about these mutations, we may learn they increase the risk for other types of cancers. Your genetic counselor will give you more information about your cancer risk if you have a mutation.

To learn more, read *Lynch Syndrome & Genetic Testing* (www.msk.org/genetics/lynch-syndrome).

部分数据表明，与出现其他林奇综合征基因突变（如 *MLH1*、*MSH2* 和 *EPCAM*）的人群相比，出现 *MSH6* 基因突变的人群总体患癌风险可能较低。 与您的遗传咨询师和医疗团队保持联系非常重要，因为风险相关信息可能会随着时间变化。

如果出现 *PMS2* 基因突变，应该如何应对患癌风险?

Your genetic counselor will review your results. They will talk with you about what cancer your mutation is linked to.

Your genetic counselor will also review your personal and family history of cancer and give you cancer screening recommendations. They may recommend you start having cancer screenings at a younger age or have them more often than most people. They may also suggest you get specialized screenings to help find cancer as early as possible.

Some examples of these cancer screenings include:

- Colonoscopies starting at an earlier age and more often than most people.
- An upper endoscopy (a procedure that lets your doctor see inside your stomach and small intestine).
- A urinalysis (a test to look for blood in your urine).

Your genetic counselor may also talk with you about having surgery to try to keep cancer from developing, such as:

- Surgery to remove your uterus to prevent endometrial cancer.
- Surgery to remove your ovaries to prevent ovarian cancer.

If you’ re having surgery for colon cancer or polyps, your genetic counselor may recommend you have extra colon tissue removed to prevent colon cancer.

If you decide to have surgery, talk with your genetic counselor about the right time to have it. Surgery to remove the uterus and ovaries affects fertility (your ability to have biological children). If you plan to have biological children, your genetic counselor can talk with you about your options.

Your genetic counselor will also talk with you about whether there are any other screening or prevention options that may be right for you.

PMS2 基因突变对我的血亲意味着什么？

如果您有基因突变，您的亲生父母、兄弟姐妹和子女都有 50% 的几率具有相同的突变。 您的远房亲戚也可能会面临同样的突变风险。

男性和女性在家族中遗传变异的几率相同。 只需从父母一方继承一种突变，罹患癌症的风险就会增加。

遗传咨询师将查看您的家族史，并与您讨论是否建议对您的血亲进行基因检测。

这对生育规划意味着什么？

如果您有 *PMS2* 基因突变并计划生育，有些方案可以防止您的孩子遗传该突变。 您可能想要考虑讨论这些方案，尤其是如果您和您的伴侣都有 *PMS2* 基因突变。

如果双方都有 *PMS2* 基因突变，则您的孩子有可能会在出生时患有一种称为先天性错配修复缺陷综合征 (CMMR-D) 的严重疾病。 CMMR-D 综合征会导致儿童罹患癌症的风险较高。 如您已有孩子，则他们不太可能患有 CMMR-D，因为该病通常是在出生早期确诊。 有关基因检测和生育规划的更多信息，请咨询您的遗传咨询师。

如果我没有基因突变会发生什么？

如果您没有基因突变，或者我们发现了临床意义未明变异 (VUS)，则遗传咨询师将审查您

的个人和家族癌症病史。 他们会与您讨论您应该遵循的一般癌症筛查指南。

VUS 是一种基因变异，但我们还不知道其是否与患癌症的风险较高有关。 大多数 VUS 最终都被证实属于正常变化，不会影响健康状况。

联系方式

> 如果您有任何问题或担忧，请咨询临床遗传学服务部的遗传咨询师。 您可以在周一至周五上午 9:00 至下午 5:00 致电 646-888-4050 与他们联系。

有关更多资源，请访问 www.mskcc.org/pe，在我们的虚拟图书馆中进行搜索。

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