



患者及照护者教育

# 关于林奇综合征的多基因面板检测

本信息介绍了林奇综合征的多基因面板检测。还说明了您的检测结果可能会对您和您家人产生哪些影响。

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。

这项多基因面板检测旨在检查您的 DNA 是否存在与林奇综合征有关的基因突变。林奇综合征与结肠直肠癌、子宫（子宫内膜）癌、卵巢癌和其他类型的癌症有关。

通常情况下，与林奇综合征相关的基因有助于预防癌症。这些基因的突变会导致其停止正常运作。这会提高您罹患某些类型癌症的风险。

重要的是要明白，出现林奇综合征基因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，林奇综合征患者的癌症类型和确诊年龄也会有所不同。

如需了解更多信息，请阅读 [林奇综合征与基因检测](http://www.msk.org/genetics/lynch-syndrome) ([www.msk.org/genetics/lynch-syndrome](http://www.msk.org/genetics/lynch-syndrome))。

## 什么是多基因面板检测？

我们可以 1 次检测 1 个基因的突变，也可以同时检测多个基因的突变。多基因面板检测是指我们使用同一唾液或血液样本同时检测多种基因。通过同时检测多种基因，可以更快查明癌症的遗传成因。

## 多基因面板将检测哪些基因？

我们将检测与林奇综合征相关的 5 种基因：*MLH1*、*MSH2*、*MSH6*、*PMS2* 和 *EPCAM*。

## 如果我有基因突变会发生什么？

如果您的 5 个林奇综合征基因中有 1 个发生了突变，即意味着您会患有林奇综合征。

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](http://www.msk.org/genetics/lynch-syndrome)).

## 如果出现林奇综合征基因突变，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.

## 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。

# 基因突变对我的血亲意味着什么？

我们检测的基因突变为显性遗传。这意味着，只要从父母一方遗传到一个突变，罹患癌症的风险就更高。男性和女性在家族中遗传变异的几率相同。

如果您的其中一种基因发生变异，则其他血亲也有可能患有该种疾病。您的亲生父母、兄弟姐妹和子女也有 50% 的几率出现相同突变。

您的远房亲戚也可能面临同样的突变风险。与他们分享这些信息可能会有所帮助。包括叔叔阿姨、侄子侄女和堂表兄弟姐妹。

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？

如果您有林奇综合征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。您可能需要考虑讨论这些方案，尤其是如果您和您的伴侣都有相同的林奇综合征基因突变。

如果双方都存在相同的林奇综合征基因突变，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。

# 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](http://www.mskcc.org/pe)，在我们的虚拟图书馆中进行搜索。

---

About the Multi-Gene Panel Test for Lynch Syndrome - Last updated on August 8, 2023  
Memorial Sloan Kettering Cancer Center 拥有和保留所有权利