Memorial Sloan Kettering Cancer Center

患者及照顧者教育

關於 BRIP1 基因突變

本資訊旨在說明 *BRIP1* 基因突變會對您和您的家人構成甚麼影響。

In this resource, the word “family” means people related to you by blood. They’re not related to you through marriage or adoption. We also call these family members your blood relatives.

BRIP1 基因通常有助預防癌症。 此基因的突變會導致它停止正常運作。

It is important to understand that having a mutation in this gene does not mean you will definitely develop cancer. It means that you have an increased risk of developing certain types of cancers. The type of cancer can vary among people who have mutations in this gene, even within the same family.

如果我有 *BRIP1* 突變，我罹患癌症的風險有多大？

BRIP1 突變會增加罹患卵巢癌的風險。 *BRIP1* 突變也可能會增加罹患乳腺癌的風險，但我們需要更多研究來更深入了解這種風險。

雖然 BRIP1 突變意味着罹患癌症的風險比一般人更高，但這可能

無法完全解釋您的血親為何罹患癌症。

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.

如欲了解更多資訊，請閱讀*遺傳性卵巢和子宮癌*：www.mskcc.org/genetics/ovarian-uterine-cancer

如果我有 *BRIP1* 突變，我該如何降低患癌風險？

如您身上出現突變，則遺傳學顧問將檢視相關結果以及您本人和家族的癌症病史，然後給您建議。

他們可能會討論是否進行手術來切除卵巢，藉以預防卵巢癌。 如果您決定進行手術，請與遺傳學顧問討論適合的手術時間。 卵巢切除手術會影響生育能力（生育親生子女的能力）。 如您計劃生育親生子女，則遺傳學顧問可與您就相關選擇作討論。

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

BRIP1 突變對我的血親而言意味著甚麼？

If you have a mutation, your biological parents, siblings, and children each have a 50% chance of having the same mutation. Your distant family members may also be at risk for having the same mutation.

Males and females have an equal chance of passing down a mutation in their family. You only need to inherit a mutation from one parent to have an increased risk for cancer.

Your genetic counselor will review your family history and talk with you about whether they recommend genetic testing for your blood relatives.

這對計劃生育又意味著甚麼？

如果您有 *BRIP1* 突變並計劃生育，則有些選項可以防止孩子遺傳到該突變。 您可能需要考慮討論這些選項，特別是如果您和伴侶都有 *BRIP1* 突變。

如果你們倆都有 *BRIP1* 基因突變（這種情況很罕見），那麼您的孩子有可能出生時會患有一種稱為范康尼貧血 (FA) 的嚴重疾病。 FA 是一種遺傳性疾病，可導致先天性障礙、骨髓衰竭和癌症風險。 如您已有孩子，則他們不太可能患有 FA，因為這種疾病通常是在出生初期便被診斷出來的。 如需更多有關基因檢測和計劃生育的資訊，請向遺傳學顧問查詢。

What happens if I do not have a mutation?

If you do not have a mutation or if we find a variant of uncertain significance (VUS), your genetic counselor will review your personal and family history of cancer. They’ ll talk with you about the general cancer screening guidelines you should follow.

A VUS is a change in a gene, but we don’ t yet know if it is linked with a higher risk for cancer. Most VUS are eventually found to be normal changes that do not affect your health.

Contact information

If you have any questions or concerns, talk with a genetic counselor in the Clinical Genetics Service. You can reach them Monday through Friday from 9 a.m. to 5 p.m. at 646-888-4050.

如欲獲取更多資源，請瀏覽 www.mskcc.org/pe，於我們的虛擬圖書館內搜尋。