

Additional Findings 附帶發現

As part of the consent in joining the Hong Kong Genome Project (HKGP), participants would be asked if they would like to learn about additional genomic information that may be related to genetic diseases and health risks. This additional information is called the “Additional Findings”, in which the identified variant is disease-causing but is not related to the participants’ primary health concerns. If participants consent to receive Additional Findings, we will inform participants whether pathogenic/likely pathogenic variants are identified in the following genes.

在香港基因組計劃（計劃）的同意書內，參加者可選擇是否接收其他可能與遺傳疾病和健康風險有關的資訊。這些資訊稱為「附帶發現」（Additional Findings），顯示與參加者的主要病況無關但具致病性的基因變異。如參加者同意接收附帶發現，我們會根據以下列表，報告有關具致病性（pathogenic）/ 可能具致病性（likely pathogenic）的基因變異。

Gene List of Additional Findings 附帶發現基因列表

Diseases 疾病	Genes 基因	Adults 成人	Children 兒童
Lynch syndrome 連氏綜合症 (舊稱: 遺傳性非癌肉大腸癌綜合症)	<i>MLH1</i>	✓	
	<i>MSH2</i>	✓	
	<i>MSH6</i>	✓	
Familial adenomatous polyposis 家族性大腸腺癌肉病	<i>APC</i>	✓	✓
MYH-associated polyposis MYH 相關癌肉病	<i>MUTYH</i>	✓	
Hereditary Breast and Ovarian Cancer Syndrome 遺傳性乳癌與卵巢癌綜合症	<i>BRCA1</i>	✓	
	<i>BRCA2</i>	✓	
Von Hippel-Lindau syndrome 逢希伯-林道綜合症	<i>VHL</i>	✓	✓
Multiple endocrine neoplasia 多發性內分泌腫瘤	<i>MEN1</i>	✓	✓
	<i>RET</i>	✓	✓
Familial hypercholesterolaemia 家族性高膽固醇血症	<i>LDLR</i>	✓	✓
	<i>APOB</i>	✓	✓
	<i>PCSK9</i>	✓	✓