ACT Risk™

遺癌檢™ 癌症基因檢測

GENETIC TEST FOR HEREDITARY CANCERS

ACT GENOMICS®

9種遺傳性癌症、11種癌症症候群
67個基因一次評估！

行動基因 ACTRisk™ 透過國際資料庫GnomAD、1000 Genome、Clinvar及COSMIC、台灣資料庫Taiwan Biobank及行動基因自建亞洲人種資料庫，針對國際常見的9種遺傳性癌症(含乳癌、大腸癌、黑色素癌、胰臟癌、神經內分泌腫瘤等)、11種遺傳性癌症症候群之好發基因進行檢測，希望協助更多家庭提早評估家族癌症風險。

檢測基因	遺傳性癌症										遺傳性癌症症候群										
	乳癌	卵巢癌	大腸直腸癌	子宮內膜癌	黑色素癌	胰臟癌	胃癌	前列腺癌	神經內分泌瘤	其他癌症	Hereditary Breast and Ovarian Cancer Syndrome	Hereditary Diffuse Gastric Cancer	Lynch Syndrome	Juvenile Polyposis Syndrome	Peutz-Jeghers Syndrome	Familial Adenomatous Polyposis	MUTYH-associated Polyposis	Li-Fraumeni Syndrome	Cowden Syndrome	Melanoma-Pancreatic Cancer Syndrome	von Hippel-Lindau
APC			●			●	●			●						●					
BRCA1	●	●				●		●			●										
BRCA2	●	●			●	●		●			●										
CDH1	●						●					●									
CHEK2	●		●					●													
EPCAM		●	●	●		●	●						●								
MLH1	●	●	●	●		●	●	●					●								
MSH2		●	●	●			●	●					●								
MSH6		●	●	●			●	●					●								
MUTYH			●														●				
PALB2	●	●				●		●													
PMS2			●	●									●								
PTEN	●		●	●						●									●		
RAD50	●	●																			
RAD51C	●	●						●													
RAD51D	●	●						●													
TP53	●		●	●	●	●	●	●		●								●			
VHL									●												●
ALK										●											
ATM	●	●				●		●													
ATR								●													
AXIN2			●																		
BARD1	●																				
BLM			●																		
BMPR1A			●				●							●							
BRIP1	●	●						●													
CDK4					●																
CDKN2A					●	●														●	
CFTR						●															
ENG									●												
EPAS1									●												
FAM175A								●													
FANCC	●	●																			
FH									●												
FLCN										●											
GALNT12			●																		
GEN1								●													
GREM1			●																		
MAX									●												
MC1R					●																
MDH2									●												
MEN1									●												
MET										●											
MRE11	●	●						●													
MSH3			●																		
NBN	●	●						●													
NF1	●									●											
NF2										●											
NTHL1			●																		
POLD1			●																		
POLE			●																		
PRSS1						●															
RB1										●											
RET									●	●											
SCG5			●																		
SDHA							●		●	●											
SDHB							●		●	●											
SDHC							●		●	●											
SDHD							●		●	●											
SDHAF2									●												
SMAD4			●				●		●					●							
SPINK1						●															
STK11	●	●	●	●		●	●			●					●						
TMEM127									●												
TSC1										●											
TSC2										●											
XRCC2	●	●																			

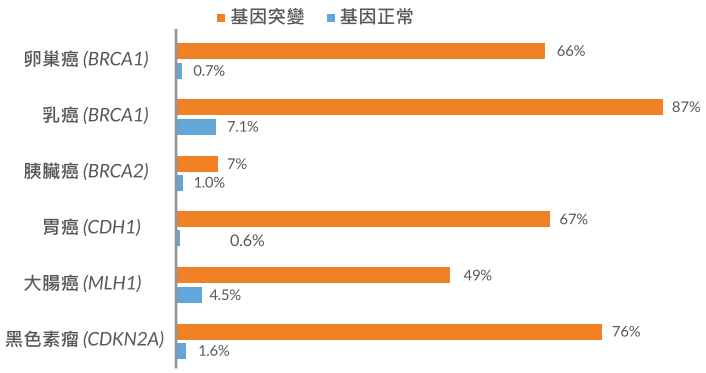
黃色框所列基因含LGR(大片段基因重組導致的缺失或重複)檢測
本基因列表僅供參考，行動基因保有最後檢測基因決定權

了解67個癌症基因 規劃家族醫療計畫

當同一家族出現多位癌症病患時，需考慮家庭成員是否暴露在相似危險因子中，如吸煙、致癌物等；或者，應該考慮遺傳基因變異問題。

什麼是遺傳性癌症

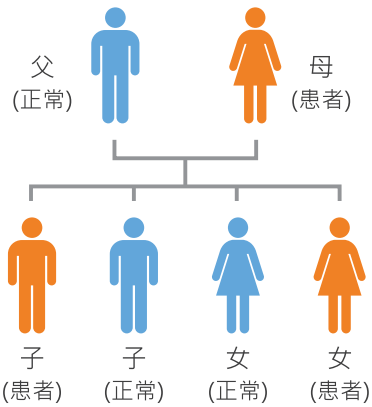
癌症患者中有5-10%是由遺傳性基因變異所引起[1]，除了眾所熟知的*BRCA1/2*基因變異會導致乳癌、卵巢癌、攝護腺癌等多種癌症的罹癌風險提高之外，目前已知有多種癌症或症候群與相關基因變異有關，帶有基因變異者的罹癌風險比一般大眾高出10~90倍之多（圖一）[2~5]。



圖一、癌症基因變異導致罹癌風險上升

雖然遺傳性癌症僅在所有癌症病患中占一小部分，但仍不能忽視。根據2005年一針對33,197名癌症患者中的大型研究發現，有14.6% 具中度家族癌症的病史，其罹癌風險較一般家族提升2倍；而有 7.7%的患者，具有強烈的家族癌症病史，罹癌風險則提升5-7倍[6]。

這類的家族中多帶有癌症易感基因(Cancer Susceptibility Gene/Predisposing Gene)，所謂的癌症易感基因指的是可以依照癌症的外顯率(Penetrance)分成高風險(High Penetrance，如*TP53*、*BRCA1*、*BRCA2*與*PTEN*等)、中度風險(Moderate Penetrance，如*RAD51C*、*CHEK2*等)與低風險(Low Penetrance)基因[7]。帶有中度到高度風險基因的患者，其罹癌風險顯著上升。因此依照美國臨床腫瘤醫學會與NCCN guideline的建議，需要及早針對特定器官進行癌症篩檢、監測及介入。2017年即有研究指出，帶有大腸癌易感基因突變的患者，若定期進行大腸鏡檢查及瘜肉切除，相較於未篩檢的組別，可降低大腸癌的發生率，並因為早期介入，使死亡率下降65%[7]。



圖二、*BRCA1*基因自體顯性遺傳

癌症的遺傳模式

一般來說，遺傳性癌症患者通常有家族癌症病史，父系與母系皆須考慮，而且未必是相同癌症。以乳癌患者常見的*BRCA1/2*基因突變為例，皆為自體顯性遺傳（圖二），因此遺傳性乳癌/卵巢癌症候群中，其家族子代中所有成員皆有50%之機會帶有突變基因。如果母親*BRCA*基因變異，則後代不論男女有50%的機會遺傳到該突變基因。同時，家族中罹患卵巢癌、胰臟癌、攝護腺癌的比例也相對地較高，且癌發年紀與同種癌症的平均發生年紀相比，有可能較為年輕。由於每種遺傳性癌症的特徵不盡相同，執行檢驗前，建議先進行遺傳諮詢(Genetic Consulting)。

1. NIH, Genetic Testing for Inherited Cancer Susceptibility Syndromes
2. Kuchenbaecker, KB, et al. JAMA. 2017;317(23):2402–2416.
3. NCCN Guidelines: Genetic/Familial High-Risk Assessment: Colorectal (2019. V3)
4. NCCN Guidelines: Genetic/Familial High-Risk Assessment: Breast and Ovarian (2020. V1)
5. NCCN Guidelines: Gastric Cancer (2017. V3)
6. Semin Oncol. 2016 Oct;43(5):528-535
7. Emma Steel, et al. Hered Cancer Clin Pract. 2017;15:1

ACTRisk™ 完整檢測報告

ACTRisk™ Report

Identifier
Project ID:
Report No.:
Report Date:

Subject		
Identifier:		Subject ID:
Date of Birth:		Gender:
Diagnosis:		
Ordering Physician		
Referral Doctor:		Tel:
Referral Institution:		
Address:		
Specimen		
Specimen ID:	Collection Site:	Specimen Type:
Date Received:	Sample ID:	D/ID:

ABOUT ACTRisk™

ACTRisk™ is a next-generation sequencing (NGS) assay profiling 67 genes associated with hereditary cancer. For further details of the test, please refer to the "TEST DETAILS" section.

Testing Results of Variants/Biomarkers with Clinical Relevance

Pathogenic/Likely Pathogenic Variants: Positive			
Genomic Alterations	**Transcript**	**Zygosity**	**Classification**
BRCA2 c.994del (I332fs)	NM_000059	Heterozygous	Pathogenic

Cancer Risk Evaluation		
***BRCA2* Mutation Carrier**		
Cancer	**Risk**	**General Population Risk**
Breast	55-69%	13.0%
Ovarian	13-29%	1.3%
Prostate	19-61%	11.6%
Pancreatic	5-10%	1.5%

*NCCN guidelines: Genetic/Familial High-Risk Assessment: Breast, Ovarian, Pancreatic, and Prostate (2025. V2)

Management Recommendation			
***BRCA2* Mutation Carrier**			
Cancer	**Management**	**Starting Age**	**Frequency**
Breast (Female)	Risk-reducing mastectomy and Risk-reducing agents	Discuss with doctor	NA
	MRI or Mammogram	30-75 year-old	Annually
	Clinical breast examination	25 year-old	Every 6-12 months

ACTRisk™ Report

Identifier
Project ID:
Report No.:
Report Date:

Variant Interpretation

BRCA2 I332fs	**Clinical Significance**
	Sequencing analysis confirmed the presence of the heterozygous germline sequence change in BRCA2 c.994del (I332fs). This mutation is predicted to result in the formation of a premature stop codon. At the time of original testing, the variant meets the ACMG[1] criteria to be classified as a pathogenic variant, with pathogenic criterion weighted as very strong (PVS1). The variant detected in BRCA2 has been classified as associated with an increased risk for the Hereditary Breast and Ovarian Cancer Syndrome. As this is a germline mutation, the patient also has an increased risk of developing a wide variety of other cancers, including breast, ovarian, prostate and pancreatic. Estimated until 70 years old, the lifetime risk for breast cancer is 55-69%, for ovarian cancer is 13-29%, for prostate cancer is up to 19-61%, for pancreatic cancer is 5-10%.
	Biological Impact
	The BRCA2 gene encodes a tumor suppressor involved in the homologous recombination pathway for double-strand DNA repair[2]. BRCA2 has been implicated as a haploinsufficient gene with one copy loss may lead to weak protein expression and is insufficient to execute its original physiological functions[3]. BRCA2 germline mutations confer an increased lifetime risk of developing breast, ovarian, prostate and pancreatic cancer, limited reports of related gastric cancer, and Fanconi anemia subtype D1-associated risk of brain cancer, medulloblastoma, pharyngeal cancer, chronic lymphocytic leukemia and acute myeloid leukemia[4]. Somatic mutations in BRCA2 are highest in colorectal, non-small cell lung cancer (NSCLC), and ovarian cancers[5].

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ACT Genomics laboratory is accredited by CAP (CAP number: X030901).
ACT Genomics only provides a technical report of the test; please consult a specialist physician to determine the appropriate clinical actions and follow the instructions of the physician. The results are only valid for the tested sample(s).
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ACTRisk™ Report

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Project ID:
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Ovarian	Risk-reducing agents	Discuss with doctor	NA
	Serum CA-125 and pelvic ultrasound	Individualized	NA
	Risk-reducing salpingo-oophorectomy	40-45 year-old	NA
Breast (Male)	Clinical breast examination	35 year-old	Annually
	Breast self-exam and education	35 year-old	Individualized
Prostate	Prostate cancer screening	40 year-old	Individualized
Pancreatic	Consider MRI/MRCP or Endoscopic ultrasonography screening	50 year-old	Annually

*NCCN guidelines: Genetic/Familial High-Risk Assessment: Breast, Ovarian, Pancreatic, and Prostate (2025. V2)

Variants of Unknown Clinical Significance			
Genomic Alterations	**Transcript**	**Zygosity**	**Classification**
ALK c.3574C>G (R1192G)	NM_004304	Heterozygous	VUS
APC c.7061C>T (A2354V)	NM_000038	Heterozygous	VUS

About the Interpretation of the ACTRisk™

- Genetic counseling should be offered to patients to discuss the clinical implications of the test result. Any interpretation provided in this report should clinically correlate with the patient's profile and relevant family history. Genetic counseling should only be provided by healthcare professionals with relevant qualifications or training.
- Patients should consider sharing their test result with other family members if the result is positive. Depending on the mode of inheritance, each of the siblings or children of the patient may have up to 50% chance of inheriting the same mutation. Germline testing is not recommended in minors as the test result may not benefit individuals in early childhood.
- Follow-up testing of the identified germline mutation in other high-risk family members of the patient may help them benefit from surveillance and early intervention to mitigate or lower their cancer risk.
- The information provided in the test report is not intended as a substitute for medical advice. Please consult with a physician or other healthcare professionals should any questions arise.
- VUS variants have unknown effects on gene function, have not been previously reported or have been reported with inadequate or conflicting evidence regarding pathogenicity, clinical relevance, or cancer risk. Therefore, variants classified as VUS should not be used in clinical decision making.
- Please note that low penetrance and late age-of-onset variants that are associated with diseases may be present at a low frequency in large population studies.

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基因位點突變 SNV & INDELS

DNA序列中的核苷酸(Nucleotide)產生位點突變時，可能造成致癌基因不受調控而持續活化，或造成抑癌基因失去功能。

大片段基因重組 LGR

提供大片段基因重組(Large Genomic Rearrangements)導致的缺失(Deletion)或重複(Duplication)。

癌症風險與管理資訊 Cancer Risk & Management

針對檢測結果，提供癌症後續風險評估及疾病管控建議。

註1：行動基因所有檢測皆於CAP合格認證實驗室進行檢測，並依結果出具技術檢測報告。關於報告的臨床解釋及相關醫療問題，請向專業醫師諮詢相關建議。以上圖像皆為說明用途，實際圖像可能與上述有所不同。

註2：由於檢測技術限制以及個體腫瘤基因差異等原因，即使檢測人員已確實執行標準操作程序，仍可能在檢測特定基因上未發現任何突變異常。

ACTRisk™ 檢測規格

檢體需求
血液 4~8ml

檢測時間
12個工作天
*以認證實驗室收到合格檢體始算

檢測基因數
以NGS全面檢測67個遺傳性癌症相關基因

分析變異種類
-單核苷酸變異(SNV)
-小片段插入/缺失(Small InDel)
-基因特定剪接位點突變(Splicing Site Mutation)
-18個關鍵基因大片段基因重組(LGR, Large Genomic Rearrangements)，如*BRCA1/2*、*MMR* Genes

採用資料庫
報告分析採用國際資料庫GnomAD、1000Genome、Clinvar及COSMIC、台灣資料庫Taiwan Biobank及行動基因自建亞洲人種資料庫，佐證詳細

建議檢測家族

ACTRisk™ 檢測優勢

67個遺傳性癌症基因相關資訊
抽血一次即可檢測，利用NGS進行67個基因全外顯子定序，全面評估9種重大遺傳性癌症、11種癌症症候群基因。

國際資料庫提供完整資訊
國際資料庫GnomAD、1000 Genome、Clinvar及COSMIC、台灣資料庫Taiwan Biobank及行動基因自建亞洲人種資料庫。

NGS高準確度檢測
不僅僅只分析突變熱點，更涵蓋SNV、Small InDel、Splicing Site和LGR等各類型突變。

詳細醫學報告
提供最新國際臨床研究文獻相關風險資訊與NCCN建議相應臨床處置，完整資訊，清楚易懂。

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

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