Elite 菁英診所 CLINIC

報告編號：NGS*******R

金萬林實驗室

新北市汐止區新台五路 1 段 97 號 30 樓

TEL: +02-2697-6888, FAX: +886-2-2697-6777

廣泛型癌症循環核酸檢測報告書

送檢資料 [1]

送檢單位： ****/**	**送檢醫師：** *** 醫師
性　　別： ☐ 男　■ 女	**聯絡電話：** **-*******
檢體編號： *******	**電子郵件：** ****@**.***.** (用以寄送檢測報告電子檔)
檢測流水號： NGS*******	**採檢日期(YYYY/MM/DD)：** ****/**/**

檢體種類： ■ 血液

根據該樣品「檢測申請書/檢測同意書」所述，

「運送條件及樣本質量請參閱送檢須知，若不符合允收標準或檢測標準是否仍要進行檢測？

☐ 是。仍要進行檢測。*(本實驗室保留最終解釋權。)*

■ 否。予以退件。」

檢體品質 [2,3,4]

收檢品質： ■ 檢體**符合**允收標準。

☐ 檢體量**未達**允收標準。

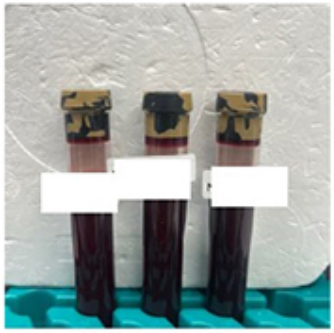
☐ 檢體運送條件**未達**允收標準。

檢測品質： ■ 檢體**符合**檢測標準。

☐ 檢體 DNA 去氧核醣核酸濃度**未達**檢測標準。

☐ 檢體次世代定序平均深度**未達**檢測標準。

檢體照片：



金萬林實驗室
新北市汐止區新台五路 1 段 97 號 30 樓
TEL: +02-2697-6888, FAX: +886-2-2697-6777

Gene Testing Report[5,6]

檢測項目：廣泛型癌症循環核酸檢測	報告編號：NGS*******R
收件時間(YYYY/MM/DD)：****/**/**	檢測區間(YYYY/MM/DD)：****/**/**-****/**/**
報告時間(YYYY/MM/DD)：****/**/**	檢測週期：10 工作天
檢測技術背景 (Testing method)	
廣泛型癌症循環核酸檢測係利用標的擴增方式(target amplification)，分析多個癌症相關基因變異狀態。此檢測於單一工作流程下，使用次世代定序方法檢測 46 個基因之變異，包含單點變異(Single Nucleotide Variant, SNV)與片段插入或缺失(insertion or deletion, indel)。	
檢測侷限 (Testing limitations)	
1. 單點變異與片段插入或缺失的變異位點頻率(allele frequency)偵測極限為 0.5%。 2. 當送檢樣本未符合允收標準，可能導致檢測不確定性(Uncertainty)。	
檢測基因 (Testing genes)	
SNV and indel: AKT1, ALK, APC, AR, ARAF, BRAF, CHEK2, CTNNB1, DDR2, EGFR, ERBB2, ERBB3, ESR1, FBXW7, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNA11, GNAQ, GNAS, HRAS, IDH1, IDH2, KIT, KRAS, MAP2K1, MAP2K2, MET, MTOR, NRAS, NTRK1, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, SF3B1, SMAD4, SMO, TP53	

核酸品質[3]

DNA 去氧核醣核酸濃度：12.7 ng/μl	■ DNA 去氧核糖核酸濃度**符合**檢測標準
	□ DNA 去氧核糖核酸濃度**未達**檢測標準

註：檢測標準：DNA 去氧核醣核酸濃度 ≥ 1 ng/μl

定序品質[4]

次世代定序平均深度：83,815 倍	■ 定序平均深度**符合**檢測標準
	□ 定序平均深度**未達**檢測標準

註：檢測標準：次世代定序平均深度 ≥ 20,000 倍

金萬林實驗室
新北市汐止區新台五路 1 段 97 號 30 樓
TEL: +02-2697-6888, FAX: +886-2-2697-6777

重要單點變異與片段插入或缺失 [7,8,9,10,11]

基因名稱 Gene	變異類型 Alteration	核酸變異 Mutation	胺基酸變異 Amino acid change	位點深度 Depth	變異位點頻率 Allele frequency	變異位點闡述 Variant interpretation
KRAS	SNV	c.35G>A	p.(G12D)	82,637	2.40%	● Pathogenic

未檢出變異位點 [10]

SNV and indel:
AKT1, ALK, APC, AR, ARAF, BRAF, CHEK2, CTNNB1, DDR2, EGFR, ERBB2, ERBB3, ERG, ESR1, FBXW7, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNA11, GNAQ, GNAS, HRAS, IDH1, IDH2, KIT, MAP2K1, MAP2K2, MET, MTOR, MYC, NRAS, NTRK1, NTRK3, PIK3CA, PTEN, RAF1, RET, ROS1, SF3B1, SMAD4, SMO, TP53

檢測人員	報告簽署
[redacted]	[redacted]

金萬林實驗室
新北市汐止區新台五路 1 段 97 號 30 樓
TEL: +02-2697-6888, FAX: +886-2-2697-6777

Sample Cancer Type: Lung Cancer

Variant Details

DNA Sequence Variants

Gene	Amino Acid Change	Coding	Locus	Allele Frequency	Transcript	Variant Effect	Oncomine Variant Class
KRAS	p.(G12C)	c.34G>T	chr12:25398285	21.52%	NM_033360.4	missense	Hotspot
PDGFRA	p.(P567=)	c.1701A>G	chr4:55141055	100.00%	NM_006206.6	synonymous	
TP53	p.(H193L)	c.578A>T	chr17:7578271	23.99%	NM_000546.6	missense	

金萬林實驗室
新北市汐止區新台五路 1 段 97 號 30 樓
TEL: +02-2697-6888, FAX: +886-2-2697-6777

Biomarker Descriptions

KRAS G12C

KRAS proto-oncogene, GTPase

Background: The KRAS proto-oncogene encodes a GTPase that functions in signal transduction and is a member of the RAS superfamily which also includes NRAS and HRAS. RAS proteins mediate the transmission of growth signals from the cell surface to the nucleus via the PI3K/AKT/MTOR and RAS/RAF/MEK/ERK pathways, which regulate cell division, differentiation, and survival[1,2,3].

Alterations and prevalence: Recurrent mutations in RAS oncogenes cause constitutive activation and are found in 20-30% of cancers. KRAS mutations are observed in up to 10-20% of uterine cancer, 30-35% of lung adenocarcinoma and colorectal cancer, and about 60% of pancreatic cancer[4]. The majority of KRAS mutations consist of point mutations occurring at G12, G13, and Q61[4,5,6]. Mutations at A59, K117, and A146 have also been observed but are less frequent[7,8].

Potential relevance: The FDA has approved the small molecule inhibitors, sotorasib[9] (2021) and adagrasib[10] (2022), for the treatment of adult patients with KRAS G12C-mutated locally advanced or metastatic non-small cell lung cancer (NSCLC). Sotorasib and adagrasib are also useful in certain circumstances for KRAS G12C-mutated pancreatic adenocarcinoma[11]. The FDA has also granted breakthrough therapy designation (2022) to the KRAS G12C inhibitor, GDC-6036[12], for KRAS G12C-mutated non-small cell lung cancer. The SHP2 inhibitor, BBP-398[13] was granted fast track designation (2022) in combination with sotorasib for previously treated patients with KRAS G12C-mutated metastatic NSCLC. The RAF/MEK clamp, avutometinib[14] was also granted fast track designation (2024) in combination with sotorasib for KRAS G12C-mutated metastatic NSCLC who have received at least one prior systemic therapy and have not been previously treated with a KRAS G12C inhibitor. The PLK1 inhibitor, onvansertib[15], was granted fast track designation (2020) in combination with bevacizumab and FOLFIRI for second-line treatment of patients with KRAS-mutated metastatic colorectal cancer (mCRC). The EGFR antagonists, cetuximab[16] and panitumumab[17], are contraindicated for treatment of colorectal cancer patients with KRAS mutations in exon 2 (codons 12 and 13), exon 3 (codons 59 and 61), and exon 4 (codons 117 and 146)[8]. Additionally, KRAS mutations are associated with poor prognosis in NSCLC[18].