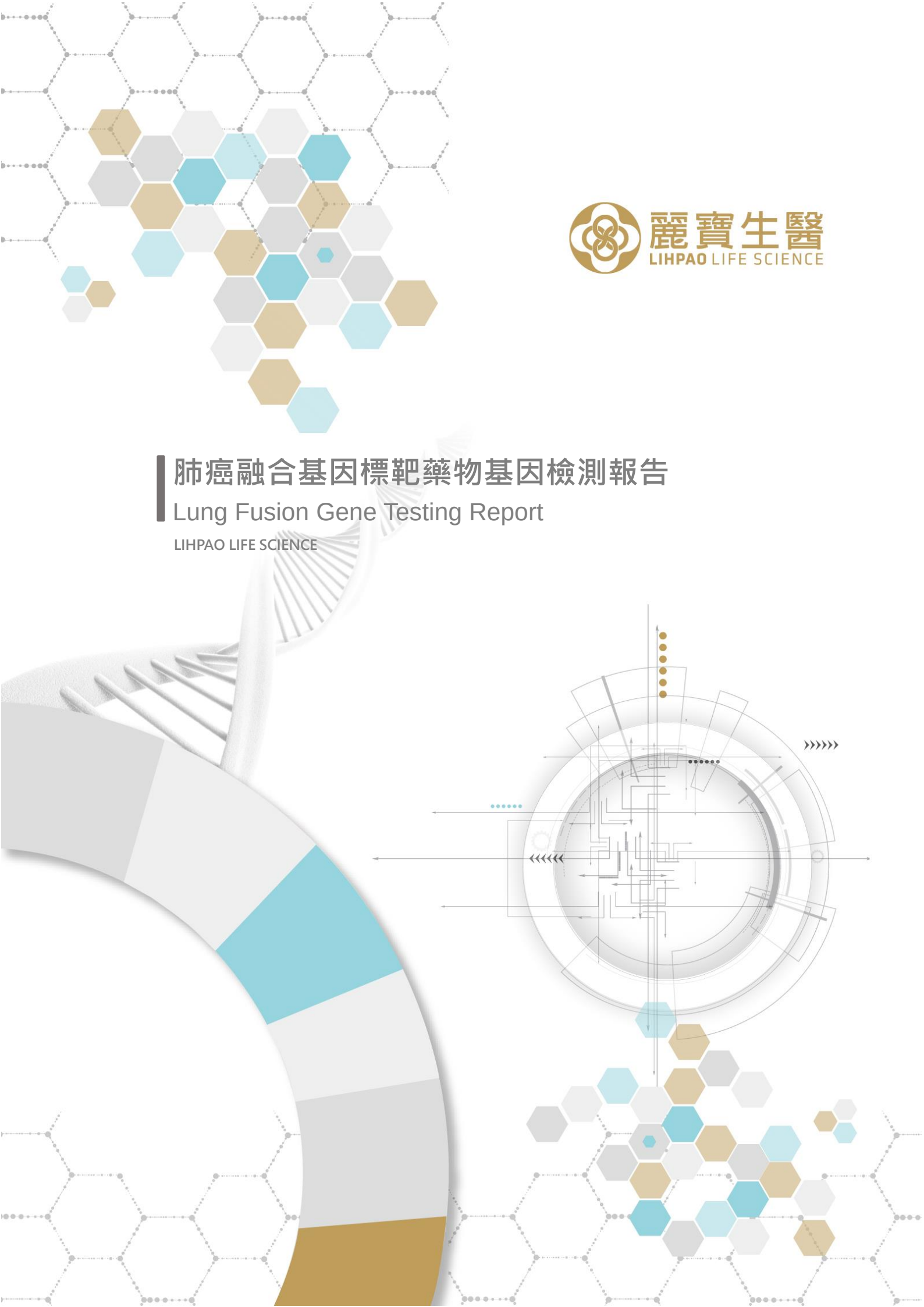
麗寶生醫
LIHPAO LIFE SCIENCE

肺癌融合基因標靶藥物基因檢測報告

Lung Fusion Gene Testing Report

LIHPAO LIFE SCIENCE



個案資料 Case Information

ORDER INFORMATION

Lung Fusion Gene Testing Report

報告編號 (Record No.)	：	YL23_029
檢體類型 (Sample Type)	：	石蠟包埋組織切片 (FFPE)
檢體編號 (Sample No.)	：	S11204465
病例號碼 (Chart No.)	：	0006023778
採集日期 (Collecting Date)	：	2023.06.07
收檢日期 (Receiving Date)	：	2023.06.08
結果日期 (Reporting Date)	：	2023.06.17
委託機構 (Referral Agency)	：	光田綜合醫院
醫師 (Physician)	：	郭集慶
聯絡人 (Contact Person)	：	郭集慶
電話 (Phone)	：	N/A
信箱	：	N/A

薛羽倢 2024-01-24

Max Cheng 2024-01-26

個案資料 Case Information		
報告編號 (Record No.): YL23_029	檢體編號 (Sample No.): S11204465	病例號碼 (Chart No.): 0006023778
委託機構 (Referral Agency): 光田綜合醫院		

肺癌融合基因標靶藥物基因檢測結果摘要

本次檢測未檢測到肺癌融合基因。

In this testing report, no lung fusion gene was detected.

樣本品質備註：

樣本品質需符合品管標準方得執行後續檢測，除特別標註外，皆代表樣本通過品管要求。

報告簽署人：

薛羽倢 2023-06-29

諮詢醫師註解：

諮詢醫師 / 日期

薛羽倢 2023-06-29

（奇美醫院精準醫療實驗室）
病解專醫字 382 號

個案資料 Case Information		
報告編號 (Record No.): YL23_029	檢體編號 (Sample No.): S11204465	病例號碼 (Chart No.): 0006023778
委託機構 (Referral Agency): 光田綜合醫院		

檢測結果 Testing Results

No.	Fusion Type	Partner Gene	Target Gene	Read Count
未檢測到與肺癌相關的融合基因 No lung fusion gene was detected in this testing				

檢測結果限制 Testing Result Limitations:

1. 本定序方式僅針對 *ALK*、*NTRK1*、*RET*、*ROS1* 四個肺癌相關的融合基因進行偵測。
   The sequencing only tests four lung fusion genes *ALK, NTRK1, RET,* and *ROS1*.
2. 本檢測報告僅供研究及醫師臨床參考，其疾病之診斷仍需配合臨床症狀。
   This report serves as reference for research and clinical purpose only, and its interpretation shall be made in conjunction with clinical symptoms by physicians.

個案資料 Case Information		
報告編號 (Record No.): YL23_029	檢體編號 (Sample No.): S11204465	病例號碼 (Chart No.): 0006023778
委託機構 (Referral Agency): 光田綜合醫院		

用藥資訊 Drug Information

No.	Variant	Recommended*	Not recommended*
本檢測結果無 FDA 核可或 NCCN 建議用藥 There are no related FDA approved or NCCN recommended therapies			

* 藥物資訊依據美國國家癌症資訊網指引或經美國食藥署認證。
The drug information is based on National Comprehensive Cancer Network guideline or approved FDA therapies.

免責聲明 Disclaimer:

1. 此報告所列之基因，其對應之潛在臨床效益或缺乏潛在臨床效益的用藥並無建議使用順序。
   In this report, the finding of any gene and any drug with a potential clinical benefit or lack of clinical benefit are NOT recommended in any specified order.
2. 此報告不保證任何特定用藥對於病患具有明顯治療效果，且缺乏潛在臨床效益的用藥同樣不保證不具有治療效果。
   This report does NOT guarantee that a specific drug will be effective in the treatment of disease in every patient. In addition, we cannot promise that a drug that lacks of clinical benefit will provide no clinical benefit.
3. 此報告中提及之用藥不適用於所有病患，所有治療皆由醫師自行決定，而其決定不應該基於報告之任何單一檢測結果與資訊。在醫師推薦病患療程前，應將本報告之資訊與病患過往病歷資料共同納入考量。
   Drugs referred in this report may not be suitable for each patient. All the treatment decisions are made by physicians. Physicians' decisions shall not be based on a single test or the information provided in this report. The information in this report shall be considered in conjunction with all relevant information from the patient's record before his/her physician recommends any treatment.

個案資料 Case Information		
報告編號 (Record No.): YL23_029	檢體編號 (Sample No.): S11204465	病例號碼 (Chart No.): 0006023778
委託機構 (Referral Agency): 光田綜合醫院		

產品介紹 Introduction

在台灣，肺癌是致死率極高的癌症，其主要的症狀包含頻繁咳嗽、喘鳴，呼吸短促與胸痛等。除了 *EGFR* 等基因的單點核苷酸或是小片段插入缺失變異之外，染色體重組形成的融合基因也被發現是肺癌發生的成因之一，融合基因的發生主要和 DNA 的斷裂導致的染色體重新排列有關，如易位 (translocation)、中間缺失 (interstitial deletion) 或是反轉 (inversion) 等。對於肺癌病患而言，有效偵測出其是否帶有 *ALK, NTRK1, RET* 與 *ROS1* 之融合基因變異，並予以正確的精準治療是提高存活率的不二法門 [1-4]。因此，本檢測透過次世代定序技術針對 4 個肺癌融合驅動基因 (driver gene) 進行基因定序，並比對融合基因資料庫中可能帶有的伴隨基因 (partner gene)，提供專業醫師相關的用藥參考。

Lung cancer has been a leading cause of death in Taiwan, and it is characterized by coughing, wheezing, shortness of breath, and chest pain. Besides single nucleotide variants, small insertions and small deletions in genes such as *EGFR*, fusion genes resulting from chromosomal rearrangement are now reported to be another kind of factors leading to lung cancer. Gene fusion occurs when DNA breaks following by chromosomal translocation, interstitial deletion or inversion. For patients with lung cancer, detection of fusion genes may increase survival rate in specific patients by treating with targeted therapies. Hence, the genetic testing of the four fusion driver genes (*ALK, NTRK1, RET* and *ROS1*) and their partner genes using next generation sequencing can provide physicians related drug information for follow-up treatment.

個案資料 Case Information		
報告編號 (Record No.): YL23_029	檢體編號 (Sample No.): S11204465	病例號碼 (Chart No.): 0006023778
委託機構 (Referral Agency): 光田綜合醫院		

融合基因列表 Fusion Gene List

CCDC6(1)-RET(12)	*CD74(3)-NTRK1(13)*	*CD74(6)-ROS1(32)*	*CD74(6)-ROS1(34)*
CEL(7)-NTRK1(7)	*CUX1(10)-RET(12)*	*EML4(13)-ALK(20)*	*EML4(13)-ALK(20)*
EML4(13)-ALK(20)	*EML4(14)-ALK(20)*	*EML4(14)-ALK(20)*	*EML4(15)-ALK(20)*
EML4(17)-ALK(20)	*EML4(17)-ALK(20)*	*EML4(18)-ALK(20)*	*EML4(2)-ALK(20)*
EML4(2)-ALK(20)	*EML4(20)-ALK(20)*	*EML4(20)-ALK(20)*	*EML4(6)-ALK(19)*
EML4(6a)-ALK(20)	*EML4(6a)-ALK(20)*	*EML4(6b)-ALK(20)*	*EML4(6b)-ALK(20)*
EZR(10)-ROS1(34)	*GOPC(4)-ROS1(36)*	*GOPC(8)-ROS1(35)*	*HIP1(21)-ALK(20)*
HIP1(28)-ALK(20)	*IRF2BP2(1)-NTRK1(10)*	*KIF5B(15)-ALK(20)*	*SLC34A2(13)-ROS1(32)*
KIF5B(15)-RET(11)	*KIF5B(15)-RET(12)*	*KIF5B(16)-RET(12)*	*SQSTM1(5)-NTRK1(10)*
KIF5B(22)-RET(12)	*KIF5B(23)-RET(12)*	*KIF5B(24)-ALK(20)*	*NFASC(18)-NTRK1(10)*
KIF5B(24)-RET(8)	*KLC1(9)-ALK(20)*	*LRIG3(16)-ROS1(35)*	*MPRIP(14)-NTRK1(13)*
TPM3(7)-ROS1(35)	*SDC4(4)-ROS1(34)*	*KIF5B(24)-RET(11)*	*NTRK1(17)-DYNC2H1(86)*
SDC4(2)-ROS1(32)	*SDC4(2)-ROS1(34)*	*SDC4(4)-ROS1(32)*	*MPRIP(18)-NTRK1(13)*
KIF5B(17)-ALK(20)	*SLC34A2(4)-ROS1(34)*	*SLC34A2(4)-ROS1(32)*	*MPRIP(21)-NTRK1(13)*
KIF5B(15)-ALK(20)	*SSBP2(12)-NTRK1(12)*	*TFG(5)-NTRK1(10)*	*SLC34A2(13)-ROS1(34)*
TPR(15)-ALK(20)			

*括號內數字代表外顯子編號。Numbers in brackets represent exon numbers of the genes.

個案資料 Case Information		
報告編號 (Record No.): YL23_029	檢體編號 (Sample No.): S11204465	病例號碼 (Chart No.): 0006023778
委託機構 (Referral Agency): 光田綜合醫院		

檢測方法 Methodology

RNA 從 FFPE 檢體萃取出來後經過反轉錄步驟合成 cDNA 並進行文庫製備。文庫經過定量及富集後使用 NGS 平台進行定序。檢測序列分別與人類基因序列 (hg19) 或特定轉錄本比對後，再透過 Fusioncatcher [5]、Ion Reporter 等融合基因偵測工具進行分析，最後將篩選出肺癌相關的融合基因及其相關標靶用藥列於報告中。

Input RNA extracted from FFPE sample is reverse-transcribed into cDNA. Standard library preparation is conducted based on the instructions described in the manufacturer's protocol. The quantified and enriched libraries are then sequenced using NGS-based platform. The sequencing reads are then aligned to the human reference genome (hg19) or specific human gene transcripts respectively. In order to detect fusion genes, Fusioncatcher [5] and Ion Reporter are used. Finally, the fusion genes and the related targeted drugs are listed in the report.

資料庫與分析工具版本 Database and Tools

Fusioncatcher	1.33
Torrent Suite	5.12.2
Ion Reporter	5.16.0.2
STAR	2.7.10a
IGV	2.10.3

個案資料 Case Information		
報告編號 (Record No.): YL23_029	檢體編號 (Sample No.): S11204465	病例號碼 (Chart No.): 0006023778
委託機構 (Referral Agency): 光田綜合醫院		

參考文獻 References

1. Davies, K. D. et al. Identifying and targeting ROS1 gene fusions in non-small cell lung cancer. *Clin Cancer Res* **18**, 4570-4579 (2012).
2. Wong, D. W. et al. The EML4-ALK fusion gene is involved in various histologic types of lung cancers from nonsmokers with wild-type EGFR and KRAS. *Cancer* **115**, 1723-1733 (2009).
3. Vaishnavi, A. et al. Oncogenic and drug-sensitive NTRK1 rearrangements in lung cancer. *Nat Med* **19**, 1469-1472 (2013).
4. Tsuta, K. et al. RET-rearranged non-small-cell lung carcinoma: a clinicopathological and molecular analysis. *Br J Cancer* **110**, 1571-1578 (2014).
5. Nicorici, D. et al. FusionCatcher - a tool for finding somatic fusion genes in paired-end RNA-sequencing data. (2022).
6. Dobin, A. et al. STAR: ultrafast universal RNA-seq aligner. *Bioinformatics* **29**, 15-21 (2013).
7. Robinson, J. T. et al. Integrative genomics viewer. *Nat Biotechnol* **29**, 24-26 (2011).