

ORIGINAL ARTICLE

Genomic and Mutational Alterations of Lung Cancer Tissues and Pleural Effusion Samples in Zhoushan Archipelago, China

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ABSTRACT

Background: This study aimed to characterize the genetic features of lung cancer tissues and pleural effusion samples from advanced Zhoushan Archipelago patients, providing insights for genome-guided precision therapy.

Methods: Genomic profiling was conducted by collecting 12 tissues and 12 pleural effusion samples. Whole-exome sequencing was applied to the tissue samples, while targeted gene sequencing was performed on the effusion samples using next-generation sequencing technology.

Results: A total of 89.86% of the mutations in the tumor tissues from the Zhoushan cohort were single nucleotide polymorphisms (SNPs), with C > T being the most dominant single nucleotide variation (SNV) class. The top 10 mutated genes included *EGFR*, *MUC4*, *MUC18*, *TP53*, *ABCC4*, *ADGRV1*, *AGXT*, *LRRC4C*, *MAGEL2*, and *OR1F1*. In addition, 54.05% of the mutations in the pleural effusion samples exhibited SNPs, with C > T being the most dominant SNV class. The top 10 mutated genes comprised *SETD8*, *KMT2C*, *ANKRD11*, *ARID5B*, *PIK3CA*, *FCGR3A*, *RYR2*, *MED12*, *RNF43*, and *H3F3A*. The tumor tissues had a notably higher incidence of missense mutations and a lower rate of frame-shift deletions compared to the pleural effusion samples.

Conclusions: We identified the genomic profiles of lung cancer tissues and pleural effusion samples in Zhoushan Archipelago, providing valuable information for genome-guided precision therapy.
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Supplementary Data

Table S1. Demographic and clinical data of lung cancer patients for tumor tissue sequencing.

Characteristic	n
Age (mean), years	
≤ 60	8
> 60	4
Gender	
Male	5
Female	7
Tobacco smoking history	
Never	8
Current	4
Tumor location	
Upper right lobe	1
Lower right lobe	3
Upper left lobe	4
Lower left lobe	2
Right middle lung	2
Tumor size, cm	
≤ 1.0	1
> 1.0 to ≤ 2.0	5
> 2.0 to ≤ 3.0	5
> 3.0	1
Lymph node metastasis	
No	8
Yes	4
Histological subtype	
Minimally invasive adenocarcinoma	2
Predominantly acinar-like growth	3
Papillary growth	5
Wall growth	2
Pathological stage	
IA	7
IB	1
IIB	3
IIIA	1

Table S2. Demographic and clinical data of lung cancer patients for pleural effusion sequencing.

Characteristic	n
Age (mean), years	
≤ 60	2
> 60	10
Gender	
Male	7
Female	5
Tumor cell	
No	6
Yes	6
Histological subtype	
Small cell lung cancer	1
Lung adenocarcinoma	9
Squamous cell carcinoma	2
Clinical stage	
IIIB	3
IVA	9

Table S3. The details of mutation status in 12 lung cancer pleural effusion samples.

Sample	Frame-shift deletions	Frame-shift insertions	In-frame deletions	In-frame insertions	Missense mutations	Nonsense mutations	Total
S1	3	0	1	0	0	0	4
S2	3	0	1	0	11	1	16
S3	2	0	0	0	11	2	15
S4	3	0	0	0	12	1	16
S5	1	1	2	0	13	1	18
S6	6	1	0	0	9	0	16
S7	11	2	1	0	10	1	25
S8	10	1	1	0	14	1	27
S9	8	1	1	0	16	0	26
S10	16	2	3	0	8	2	31
S11	14	4	1	1	9	1	30
S12	11	3	2	0	10	3	29

Table 4. The microsatellite instability status in 12 lung cancer pleural effusion samples.

Sample	Total number of sites	Number of somatic sites	Score (%)
S1	22	1	4.55
S2	22	0	0
S3	19	0	0
S4	19	0	0
S5	19	0	0
S6	22	0	0
S7	22	0	0
S8	19	0	0
S9	19	0	0
S10	22	0	0
S11	22	0	0
S12	19	0	0