[**检测报告**] - 6901152320831094784

**基本信息**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| 姓　　名 | 王五 | 样本编号 | 1701 | 测序平台 | NextSeq&MiniSeq |
| 性　　别 | 男 | 样本类型 | FF | 取样时间 | 2019.03.25 |
| 年　　龄 | 66 | 取样方法 | 穿刺 | 收样时间 | 2019.03.26 |
| 患者编号 | NCS-201903300032-M | 取样部位 | 肺部 | 报告时间 | 2022.05.31 |
| 诊断信息 | 肺癌 | 样本来源 | 上海复旦肿瘤医院 |
| 临床信息 | 肺癌 |
| 备　　注 | 本报告中的诊断信息及临床信息来自受检者送检时提供的信息，而非来自检测结果。本检测报告不对以上信息的准确性负责。 |

**质控信息**

| **质量参数** | **质控标准** | **样本数据** | **配对数据** |
| --- | --- | --- | --- |
| 测序质量评估 | 测序总Reads数(M) | >=50.00 | 116.51 | 93.16 |
| Q20比例 | >=95.00% | 100.00% | 100.00% |
| Q30比例 | >=85.00% | 100.00% | 100.00% |
| GC比例 | 45%~48% | 46.01% | 45.99% |
| Mapping Reads数(M) | >=49.50 | 116.46 | 93.11 |
| 文库Mapping率 | >=99.00% | 99.87% | 99.86% |
| 平均插入片段长度(bp) | >=200 | 210 | 210 |
| Duplicate Reads 比例 | <=30% | 28.93% | 24.46% |
| 平均测序深度 | >=1000 | 2782 | 2373 |
| 中位数测序深度 | >=1000 | 2834 | 2419 |
| 深度>(0.2x目标测序深度)占比 | >=95% | 98.08% | 97.85% |
| 深度>(1.0x目标测序深度)占比 | >=80% | 91.56% | 89.34% |

**检测结果**

1. **SNV & INDEL 突变信息：**

| **CHROM** | **START** | **END** | **REF** | **ALT** | **GENE** | **TYPE** | **VAF** |
| --- | --- | --- | --- | --- | --- | --- | --- |
| 9 | 5065019 | 5065019 | G | C | JAK2 | SNV | 10.81% |
| 9 | 21970934 | 21970934 | G | A | CDKN2A | SNV | 3.43% |
| 9 | 21974728 | 21974728 | C | A | CDKN2A | SNV | 10.33% |
| 9 | 36882061 | 36882061 | G | T | PAX5 | SNV | 2.69% |
| 9 | 133748371 | 133748371 | C | T | ABL1 | SNV | 5.86% |
| 9 | 133760367 | 133760367 | C | A | ABL1 | SNV | 10.48% |
| 9 | 135777018 | - | - | T | TSC1 | INS | 4.81% |
| 9 | 139390791 | - | - | CC | NOTCH1 | INS | 6.97% |
| 10 | 43609712 | - | - | TATG | RET | INS | 4.39% |
| 10 | 43609715 | - | - | ACCAA | RET | INS | 3.95% |
| 10 | 43609717 | - | - | AATTAATTGATTACATTCTT | RET | INS | 3.65% |
| 10 | 89653839 | 89653839 | A | G | PTEN | SNV | 3.25% |
| 10 | 89711968 | - | - | G | PTEN | INS | 3.67% |
| 10 | 123258034 | 123258034 | A | C | FGFR2 | SNV | 3.63% |
| 11 | 533535 | 533535 | C | T | HRAS | SNV | 6.95% |
| 11 | 17112940 | 17112940 | T | C | PIK3C2A | SNV | 8.50% |
| 11 | 32414249 | - | - | CAGCTCCCCT | WT1 | INS | 9.69% |
| 11 | 64572272 | 64572272 | C | T | MEN1 | SNV | 15.65% |
| 11 | 92600168 | 92600168 | C | T | FAT3 | SNV | 3.38% |
| 11 | 108117798 | 108117798 | C | A | ATM | SNV | 21.77% |
| 11 | 108170483 | 108170486 | CTCT | - | ATM | DEL | 6.25% |
| 11 | 108214019 | 108214019 | T | A | ATM | SNV | 5.23% |
| 11 | 125513686 | 125513686 | G | A | CHEK1 | SNV | 2.04% |
| 12 | 441055 | 441055 | C | T | KDM5A | SNV | 3.29% |
| 12 | 6709009 | 6709009 | T | C | CHD4 | SNV | 2.17% |
| 12 | 25380276 | 25380276 | T | G | KRAS | SNV | 12.84% |
| 12 | 121431498 | 121431498 | G | A | HNF1A | SNV | 12.58% |
| 13 | 28964091 | 28964091 | A | G | FLT1 | SNV | 2.80% |
| 13 | 102375190 | 102375190 | C | A | ITGBL1 | SNV | 3.06% |
| 14 | 75514890 | 75514890 | G | T | MLH3 | SNV | 18.52% |
| 14 | 105246455 | 105246455 | C | T | AKT1 | SNV | 15.39% |
| 15 | 89811723 | 89811723 | T | C | FANCI | SNV | 7.45% |
| 15 | 90631838 | 90631838 | C | G | IDH2 | SNV | 6.27% |
| 15 | 93510648 | 93510648 | C | - | CHD2 | DEL | 5.74% |
| 16 | 11348964 | 11348964 | C | T | SOCS1 | SNV | 3.11% |
| 16 | 67645347 | 67645348 | AA | - | CTCF | DEL | 3.67% |
| 16 | 89805927 | 89805927 | G | A | FANCA | SNV | 13.36% |
| 17 | 7577022 | 7577022 | G | A | TP53 | SNV | 5.93% |
| 17 | 7578423 | 7578423 | C | A | TP53 | SNV | 6.96% |
| 17 | 11984671 | 11984671 | A | G | MAP2K4 | SNV | 5.91% |
| 17 | 37880996 | - | - | TTA | ERBB2 | INS | 13.21% |
| 1 | 65309780 | 65309780 | G | A | JAK1 | SNV | 2.13% |
| 1 | 115256529 | 115256530 | TG | AA | CSDE1 | Complex | 3.33% |
| 1 | 162731225 | 162731225 | T | G | DDR2 | SNV | 6.22% |
| 1 | 179095570 | 179095570 | G | A | ABL2 | SNV | 3.03% |
| 2 | 25457251 | 25457251 | T | C | DNMT3A | SNV | 3.89% |
| 2 | 25468160 | 25468160 | G | - | DNMT3A | DEL | 5.64% |
| 2 | 29416770 | 29416770 | T | A | ALK | SNV | 3.25% |
| 2 | 29420411 | 29420411 | G | T | ALK | SNV | 15.78% |
| 2 | 29432664 | 29432664 | C | T | ALK | SNV | 20.30% |
| 2 | 29443697 | 29443697 | A | G | ALK | SNV | 6.89% |
| 2 | 209113112 | 209113112 | C | A | IDH1 | SNV | 14.48% |
| 2 | 212251742 | 212251742 | C | A | ERBB4 | SNV | 4.77% |
| 3 | 10183529 | 10183545 | ATGCCCCGGAGGGCGGA | - | VHL | DEL | 9.50% |
| 3 | 10183659 | 10183659 | C | T | VHL | SNV | 4.31% |
| 3 | 10188269 | 10188282 | CCATCTCTCAATGT | - | VHL | DEL | 3.18% |
| 3 | 10191626 | 10191630 | GCACA | - | VHL | DEL | 15.67% |
| 3 | 25542806 | 25542806 | C | T | RARB | SNV | 3.11% |
| 3 | 41266107 | 41266107 | T | C | CTNNB1 | SNV | 8.37% |
| 3 | 47163586 | - | - | G | SETD2 | INS | 2.16% |
| 3 | 52443861 | 52443861 | G | T | PHF7 | SNV | 25.75% |
| 3 | 142168302 | 142168302 | T | G | ATR | SNV | 6.75% |
| 3 | 185161374 | 185161374 | G | A | MAP3K13 | SNV | 4.38% |
| 3 | 187447543 | 187447543 | C | T | BCL6 | SNV | 2.22% |
| 3 | 192125845 | 192125845 | G | A | FGF12 | SNV | 2.99% |
| 4 | 55133541 | 55133541 | T | A | PDGFRA | SNV | 17.64% |
| 4 | 55155218 | 55155218 | G | T | PDGFRA | SNV | 11.50% |
| 4 | 66356211 | 66356211 | A | G | EPHA5 | SNV | 16.06% |
| 4 | 106157446 | 106157446 | G | T | TET2 | SNV | 1.83% |
| 4 | 153245453 | 153245453 | G | C | FBXW7 | SNV | 8.41% |
| 5 | 56177964 | 56177964 | G | A | MAP3K1 | SNV | 2.23% |
| 5 | 67589147 | 67589147 | A | G | PIK3R1 | SNV | 21.44% |
| 5 | 112173983 | 112173984 | CA | - | APC | DEL | 18.80% |
| 5 | 112174579 | 112174579 | G | A | APC | SNV | 16.62% |
| 5 | 112176028 | - | - | A | APC | INS | 5.89% |
| 5 | 149495442 | 149495442 | C | T | PDGFRB | SNV | 11.07% |
| 5 | 180045774 | 180045774 | T | C | FLT4 | SNV | 7.08% |
| 6 | 106552922 | 106552922 | G | T | PRDM1 | SNV | 1.93% |
| 6 | 117679018 | 117679018 | A | G | ROS1 | SNV | 17.30% |
| 6 | 157522541 | 157522541 | G | A | ARID1B | SNV | 4.28% |
| 6 | 170844466 | 170844466 | C | G | PSMB1 | SNV | 7.57% |
| 7 | 2954933 | 2954933 | G | A | CARD11 | SNV | 8.08% |
| 7 | 26224981 | 26224981 | G | A | HNRNPA2B1 | SNV | 5.23% |
| 7 | 55219011 | 55219011 | C | T | EGFR | SNV | 13.63% |
| 7 | 55241708 | 55241708 | G | C | EGFR | SNV | 7.97% |
| 7 | 55242427 | 55242427 | C | T | EGFR | SNV | 17.33% |
| 7 | 55249012 | - | - | GGT | EGFR | INS | 16.72% |
| 7 | 55249071 | 55249071 | C | T | EGFR | SNV | 3.76% |
| 7 | 55259514 | 55259515 | CT | GA | EGFR | Complex | 2.05% |
| 7 | 55259527 | 55259527 | T | A | EGFR | SNV | 6.91% |
| 7 | 140453136 | 140453136 | A | T | BRAF | SNV | 7.91% |
| 1 | 10412757 | 10412757 | T | C | KIF1B | SNV | 2.04% |
| 17 | 37881440 | 37881440 | C | T | ERBB2 | SNV | 24.38% |
| 18 | 42530439 | 42530439 | A | C | SETBP1 | SNV | 5.19% |
| 18 | 45396908 | 45396908 | A | C | SMAD2 | SNV | 1.96% |
| 18 | 48603092 | - | - | T | SMAD4 | INS | 20.07% |
| 19 | 36223270 | 36223270 | G | A | KMT2B | SNV | 7.85% |
| 22 | 30032779 | 30032800 | GGACTCTGGGGCTCCGAGAAAC | - | NF2 | DEL | 4.73% |
| 22 | 30050666 | 30050666 | T | A | NF2 | SNV | 12.52% |

1. **CNV 突变信息：**

| **CHROM** | **START** | **END** | **GENE** | **REGION** | **TYPE** | **CN** |
| --- | --- | --- | --- | --- | --- | --- |
| 13 | 28578183 | 28674099 | FLT3 | Exon 1-25 | CNV loss | 1 |
| 8 | 128748834 | 128753209 | MYC | Exon 1-3 | CNV gain | 4 |

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| --- |
|  |
| dummyAltText |

1. **SV 突变信息：**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **CHROM1** | **BREAKPOINT1** | **GENE1** | **CHROM2** | **BREAKPOINT2** | **GENE2** | **TYPE** | **VAF** | **ANNOTATION** |
| 11 | 128675350 | FLI1 | 22 | 29684015 | EWSR1 | BND | 19.8% | FLI1-EWSR1 |
| 11 | 128675349 | FLI1 | 22 | 29684016 | EWSR1 | BND | 29.7% | FLI1-EWSR1 |
| 10 | 43609713 | RET | 10 | 32311365 | KIF5B | BND | 5.8% | RET-KIF5B |
| 10 | 43609712 | RET | 10 | 32311364 | KIF5B | BND | 8.9% | RET-KIF5B |
| 10 | 32311365 | KIF5B | 10 | 43609713 | RET | BND | 5.8% | KIF5B-RET |
| 10 | 32311364 | KIF5B | 10 | 43609712 | RET | BND | 8.9% | KIF5B-RET |