[**检测报告**] - FFPE SNV CNV SV V2.4129789358864596992

**基本信息**

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| --- | --- | --- | --- | --- | --- |
| 姓　　名 | 王五 | 样本编号 | B1701 | 测序平台 | NextSeq&MiniSeq |
| 性　　别 | 男 | 样本类型 | FF | 取样时间 | 2019-03-26 |
| 年　　龄 | 66 | 取样方法 | 穿刺 | 收样时间 | 2019-03-27 |
| 患者编号 | NCS-201903300032-M | 取样部位 | 肺部 | 报告时间 | 2020-06-20 |
| 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。  Sample 自定义字段：/opt/bwa/bwa | | | | |

**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 | 93.17 | 116.52 |
| Mapping Reads数(M) | >=49.50 | 93.12 | 116.47 |
| 文库Mapping率 | >=99.00% | 99.86% | 99.87% |
| 平均插入片段长度(bp) | >=200 | 210 | 210 |
| Duplicate Reads 比例 | <=30% | 24.47% | 28.93% |
| 平均测序深度 | >=1000 | 2373 | 2782 |
| 中位数测序深度 | >=1000 | 2420 | 2834 |
| 深度>(0.1x目标测序深度)占比 | >=95% | 98.51% | 98.61% |
| 深度>(1.0x目标测序深度)占比 | >=99% | 89.34% | 91.56% |

**检测结果**

1. **SNV & INDEL 突变信息：**

| **CHROM** | **START** | **END** | **REF** | **ALT** | **GENE** | **TYPE** | **VAF** |
| --- | --- | --- | --- | --- | --- | --- | --- |
| 3 | 192125845 | 192125845 | G | A | FGF12 | synonymous SNV | 3.00% |
| 9 | 133748371 | 133748371 | C | T | ABL1 | synonymous SNV | 5.88% |
| 9 | 133760367 | 133760367 | C | A | ABL1 | nonsynonymous SNV | 10.83% |
| 15 | 93510648 | 93510648 | C | - | CHD2 | frameshift deletion | 5.55% |
| 16 | 11348964 | 11348964 | C | T | SOCS1 | synonymous SNV | 3.12% |
| 16 | 67645347 | 67645348 | AA | - | CTCF | frameshift deletion | 3.67% |
| 16 | 89805927 | 89805927 | G | A | FANCA | synonymous SNV | 13.37% |
| 1 | 10412757 | 10412757 | T | C | KIF1B | nonsynonymous SNV | 2.01% |
| 1 | 65309780 | 65309780 | G | A | JAK1 | synonymous SNV | 2.14% |
| 1 | 115256529 | 115256530 | TG | AA | NRAS | nonframeshift substitution | 3.34% |
| 1 | 154138283 | 154138283 | G | A | TPM3 | NAN | 3.99% |
| 1 | 154138291 | 154138291 | G | A | TPM3 | NAN | 2.84% |
| 1 | 162731225 | 162731225 | T | G | DDR2 | nonsynonymous SNV | 6.35% |
| 1 | 179095570 | 179095570 | G | A | ABL2 | nonsynonymous SNV | 2.98% |
| 2 | 25457251 | 25457251 | T | C | DNMT3A | nonsynonymous SNV | 3.70% |
| 2 | 25468160 | 25468160 | G | - | DNMT3A | frameshift deletion | 5.63% |
| 2 | 29416770 | 29416770 | T | A | ALK | nonsynonymous SNV | 3.24% |
| 2 | 29420411 | 29420411 | G | T | ALK | nonsynonymous SNV | 15.65% |
| 2 | 29432664 | 29432664 | C | T | ALK | nonsynonymous SNV | 20.60% |
| 2 | 29443697 | 29443697 | A | G | ALK | nonsynonymous SNV | 6.99% |
| 2 | 209113112 | 209113112 | C | A | IDH1 | nonsynonymous SNV | 15.32% |
| 2 | 212251742 | 212251742 | C | A | ERBB4 | nonsynonymous SNV | 4.56% |
| 3 | 10183529 | 10183545 | GGAATGCCCCGGAGGGC | - | VHL | frameshift deletion | 9.68% |
| 3 | 10183659 | 10183659 | C | T | VHL | nonsynonymous SNV | 4.31% |
| 3 | 10188269 | 10188282 | CCATCTCTCAATGT | - | VHL | frameshift deletion | 2.98% |
| 3 | 10191626 | 10191630 | GCACA | - | VHL | frameshift deletion | 15.41% |
| 3 | 25542806 | 25542806 | C | T | RARB | nonsynonymous SNV | 2.74% |
| 3 | 41266107 | 41266107 | T | C | CTNNB1 | nonsynonymous SNV | 8.05% |
| 3 | 47163586 | 47163586 | - | G | SETD2 | frameshift insertion | 2.27% |
| 3 | 52443861 | 52443861 | G | T | BAP1 | nonsynonymous SNV | 25.79% |
| 3 | 52676066 | 52676066 | A | T | PBRM1 | NAN | 9.60% |
| 3 | 142168302 | 142168302 | T | G | ATR | nonsynonymous SNV | 6.95% |
| 3 | 185161374 | 185161374 | G | A | MAP3K13 | nonsynonymous SNV | 4.35% |
| 3 | 187447543 | 187447543 | C | T | BCL6 | nonsynonymous SNV | 2.10% |
| 4 | 55133541 | 55133541 | T | A | PDGFRA | nonsynonymous SNV | 16.79% |
| 4 | 55155218 | 55155218 | G | T | PDGFRA | nonsynonymous SNV | 11.20% |
| 4 | 66356211 | 66356211 | A | G | EPHA5 | nonsynonymous SNV | 15.68% |
| 4 | 106157446 | 106157446 | G | T | TET2 | stopgain | 1.82% |
| 4 | 153245453 | 153245453 | G | C | FBXW7 | nonsynonymous SNV | 8.66% |
| 4 | 153249518 | 153249518 | - | TGTCCCAC | FBXW7 | frameshift insertion | 5.21% |
| 5 | 56177964 | 56177964 | G | A | MAP3K1 | nonsynonymous SNV | 2.10% |
| 5 | 67589147 | 67589147 | A | G | PIK3R1 | nonsynonymous SNV | 20.60% |
| 5 | 112173983 | 112173984 | CA | - | APC | frameshift deletion | 18.86% |
| 5 | 112174579 | 112174579 | G | A | APC | synonymous SNV | 16.12% |
| 5 | 112176028 | 112176028 | - | A | APC | frameshift insertion | 5.52% |
| 5 | 149495442 | 149495442 | C | T | PDGFRB | nonsynonymous SNV | 11.04% |
| 5 | 180045774 | 180045774 | T | C | FLT4 | synonymous SNV | 7.35% |
| 6 | 106552922 | 106552922 | G | T | PRDM1 | nonsynonymous SNV | 1.90% |
| 6 | 114292040 | 114292040 | T | - | HDAC2 | frameshift deletion | 2.74% |
| 6 | 117679018 | 117679018 | A | G | ROS1 | nonsynonymous SNV | 16.40% |
| 6 | 157099313 | 157099315 | CAC | - | ARID1B | nonframeshift deletion | 2.80% |
| 6 | 157099333 | 157099335 | CCA | - | ARID1B | nonframeshift deletion | 2.67% |
| 6 | 157099427 | 157099429 | CAG | - | ARID1B | nonframeshift deletion | 5.56% |
| 6 | 157099982 | 157099984 | GGC | - | ARID1B | nonframeshift deletion | 3.76% |
| 6 | 157100024 | 157100026 | GGA | - | ARID1B | nonframeshift deletion | 4.85% |
| 6 | 157522541 | 157522541 | G | A | ARID1B | nonsynonymous SNV | 3.57% |
| 6 | 170844466 | 170844466 | C | G | PSMB1 | nonsynonymous SNV | 7.43% |
| 7 | 2954933 | 2954933 | G | A | CARD11 | nonsynonymous SNV | 7.97% |
| 7 | 26224981 | 26224981 | G | A | NFE2L3 | nonsynonymous SNV | 5.23% |
| 7 | 55219011 | 55219011 | C | T | EGFR | nonsynonymous SNV | 13.63% |
| 7 | 55241708 | 55241708 | G | C | EGFR | nonsynonymous SNV | 7.89% |
| 7 | 55242427 | 55242427 | C | T | EGFR | nonsynonymous SNV | 16.87% |
| 7 | 55249012 | 55249012 | - | GGT | EGFR | nonframeshift insertion | 16.81% |
| 7 | 55249071 | 55249071 | C | T | EGFR | nonsynonymous SNV | 3.79% |
| 7 | 55259514 | 55259515 | CT | GA | EGFR | nonframeshift substitution | 2.05% |
| 7 | 55259527 | 55259527 | T | A | EGFR | nonsynonymous SNV | 6.87% |
| 7 | 140453136 | 140453136 | A | T | BRAF | nonsynonymous SNV | 7.94% |
| 7 | 151932897 | 151932897 | C | A | KMT2C | NAN | 7.49% |
| 7 | 151932916 | 151932916 | C | G | KMT2C | nonsynonymous SNV | 3.57% |
| 7 | 151932923 | 151932923 | G | A | KMT2C | synonymous SNV | 3.72% |
| 7 | 151932961 | 151932961 | G | A | KMT2C | stopgain | 1.64% |
| 7 | 151935831 | 151935831 | G | A | KMT2C | synonymous SNV | 4.15% |
| 7 | 151935853 | 151935853 | T | C | KMT2C | nonsynonymous SNV | 6.11% |
| 7 | 151935910 | 151935910 | C | T | KMT2C | nonsynonymous SNV | 6.40% |
| 7 | 151945167 | 151945167 | G | T | KMT2C | synonymous SNV | 3.32% |
| 7 | 151945215 | 151945215 | T | C | KMT2C | synonymous SNV | 4.52% |
| 7 | 151945225 | 151945225 | T | C | KMT2C | nonsynonymous SNV | 3.77% |
| 9 | 5065019 | 5065019 | G | C | JAK2 | nonsynonymous SNV | 10.67% |
| 9 | 21970934 | 21970934 | G | A | CDKN2A | nonsynonymous SNV | 3.43% |
| 9 | 21974728 | 21974728 | C | A | CDKN2A | nonsynonymous SNV | 10.33% |
| 9 | 36882061 | 36882061 | G | T | PAX5 | nonsynonymous SNV | 2.66% |
| 9 | 135777018 | 135777018 | - | T | TSC1 | frameshift insertion | 4.66% |
| 9 | 139390791 | 139390791 | - | CC | NOTCH1 | frameshift insertion | 6.91% |
| 10 | 43609712 | 43609712 | - | TATG | RET | NAN | 4.37% |
| 10 | 43609715 | 43609715 | - | ACCAA | RET | NAN | 4.01% |
| 10 | 43609717 | 43609717 | - | AATTAATTGATTACATTCTT | RET | NAN | 3.72% |
| 10 | 89653839 | 89653839 | A | G | PTEN | nonsynonymous SNV | 3.09% |
| 10 | 89711968 | 89711968 | - | G | PTEN | frameshift insertion | 3.68% |
| 10 | 123258034 | 123258034 | A | C | FGFR2 | nonsynonymous SNV | 4.00% |
| 11 | 533535 | 533535 | C | T | HRAS | nonsynonymous SNV | 6.89% |
| 11 | 17112940 | 17112940 | T | C | PIK3C2A | nonsynonymous SNV | 8.74% |
| 11 | 32414249 | 32414249 | - | CAGCTCCCCT | WT1 | frameshift insertion | 9.54% |
| 11 | 64572272 | 64572272 | C | T | MEN1 | nonsynonymous SNV | 15.72% |
| 11 | 92600168 | 92600168 | C | T | FAT3 | nonsynonymous SNV | 3.38% |
| 11 | 108117798 | 108117798 | C | A | ATM | nonsynonymous SNV | 22.26% |
| 11 | 108170483 | 108170486 | TCTC | - | ATM | frameshift deletion | 6.26% |
| 11 | 108214019 | 108214019 | T | A | ATM | nonsynonymous SNV | 5.53% |
| 11 | 125513686 | 125513686 | G | A | CHEK1 | NAN | 2.01% |
| 12 | 441055 | 441055 | C | T | KDM5A | nonsynonymous SNV | 3.26% |
| 12 | 6709009 | 6709009 | T | C | CHD4 | nonsynonymous SNV | 2.02% |
| 12 | 25380276 | 25380276 | T | G | KRAS | nonsynonymous SNV | 13.13% |
| 12 | 121431498 | 121431498 | G | A | HNF1A | synonymous SNV | 12.94% |
| 13 | 25044054 | 25044054 | T | C | PARP4 | nonsynonymous SNV | 1.30% |
| 13 | 28964091 | 28964091 | A | G | FLT1 | nonsynonymous SNV | 3.24% |
| 13 | 102375190 | 102375190 | C | A | FGF14 | nonsynonymous SNV | 2.96% |
| 14 | 75514890 | 75514890 | G | T | MLH3 | nonsynonymous SNV | 18.14% |
| 14 | 105246455 | 105246455 | C | T | AKT1 | nonsynonymous SNV | 15.54% |
| 15 | 89811723 | 89811723 | T | C | FANCI | synonymous SNV | 7.63% |
| 15 | 90631838 | 90631838 | C | G | IDH2 | nonsynonymous SNV | 6.32% |
| 17 | 7577022 | 7577022 | G | A | TP53 | stopgain | 5.49% |
| 17 | 7578423 | 7578423 | C | A | TP53 | nonsynonymous SNV | 6.77% |
| 17 | 11984671 | 11984671 | A | G | MAP2K4 | NAN | 5.92% |
| 17 | 37880996 | 37880996 | - | TTA | ERBB2 | nonframeshift insertion | 13.47% |
| 17 | 37880997 | 37880997 | G | T | ERBB2 | nonsynonymous SNV | 13.47% |
| 17 | 37881440 | 37881440 | C | T | ERBB2 | nonsynonymous SNV | 24.78% |
| 17 | 41620645 | 41620648 | CTCT | - | DHX8;ETV4 | NAN | 2.39% |
| 17 | 56056587 | 56056589 | TGT | - | VEZF1 | nonframeshift deletion | 1.21% |
| 17 | 56056605 | 56056607 | TGC | - | VEZF1 | nonframeshift deletion | 3.79% |
| 18 | 42530439 | 42530439 | A | C | SETBP1 | nonsynonymous SNV | 4.88% |
| 18 | 45396908 | 45396908 | A | C | SMAD2 | nonsynonymous SNV | 2.12% |
| 18 | 48603092 | 48603092 | - | T | SMAD4 | frameshift insertion | 20.10% |
| 19 | 36223270 | 36223270 | G | A | KMT2B | synonymous SNV | 7.79% |
| 21 | 42873564 | 42873564 | G | A | TMPRSS2 | NAN | 4.19% |
| 21 | 42873568 | 42873568 | C | A | TMPRSS2 | NAN | 3.20% |
| 21 | 42873571 | 42873589 | CAACAACATTCTCCAATTG | - | TMPRSS2 | NAN | 1.52% |
| 22 | 30032779 | 30032800 | CGGACTCTGGGGCTCCGAGAAA | - | NF2 | frameshift deletion | 4.70% |
| 22 | 30050666 | 30050666 | T | A | NF2 | nonsynonymous SNV | 12.08% |

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：**

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| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **CHROM1** | **BREAKPOINT1** | **GENE1** | **CHROM2** | **BREAKPOINT2** | **GENE2** | **TYPE** | **VAF** | **ANNOTATION** |
| 10 | 32311364 | KIF5B | 10 | 43609712 | RET | INV | 8.5% | KIF5B-RET |
| 11 | 128675349 | FLI1 | 22 | 29684016 | EWSR1 | BND | 29.0% | FLI1-EWSR1 |
| 11 | 128675350 | FLI1 | 22 | 29684015 | EWSR1 | BND | 19.3% | FLI1-EWSR1 |