**Illumina-B1701-SNV\_Indel预期结果**

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Chrom** | **Start** | **End** | **Ref** | **Alt** | **Gene** | **Type** | **VAF** | **CHGVS** | **PHGVS** |
| chr1 | 10412757 | 10412757 | T | C | KIF1B | SNV | 0.02 | c.3880T>C | p.S1294P |
| chr1 | 65309780 | 65309780 | G | A | JAK1 | SNV | 0.02 | c.2370C>T | p.G790G |
| chr1 | 115256529 | 115256530 | TG | AA | NRAS | Sub | 0.03 | c.181\_182CA>TT | p.Q61L |
| chr1 | 162731225 | 162731225 | T | G | DDR2 | SNV | 0.06 | c.1080T>G | p.S360R |
| chr1 | 179095570 | 179095570 | G | A | ABL2 | SNV | 0.03 | c.521C>T | p.S174L |
| chr10 | 89653839 | 89653839 | A | G | PTEN | SNV | 0.03 | c.137A>G | p.Y46C |
| chr10 | 89711968 | 89711969 | . | G | PTEN | INS | 0.03 | c.586\_587insG | p.H196fs\*6 |
| chr10 | 123258034 | 123258034 | A | C | FGFR2 | SNV | 0.04 | c.1647T>G | p.N549K |
| chr11 | 533535 | 533535 | C | T | HRAS | SNV | 0.07 | c.368G>A | p.R123H |
| chr11 | 17112940 | 17112940 | T | C | PIK3C2A | SNV | 0.09 | c.4819A>G | p.T1607A |
| chr11 | 32414249 | 32414250 | . | CAGCTCCCCT | WT1 | INS | 0.08 | c.1097\_1098ins10 | p.S367fs\*23 |
| chr11 | 64572272 | 64572272 | C | T | MEN1 | SNV | 0.15 | c.1367G>A | p.R456H |
| chr11 | 92600168 | 92600168 | C | T | FAT3 | SNV | 0.04 | c.1645C>T | p.R549W |
| chr11 | 108117798 | 108117798 | C | A | ATM | SNV | 0.22 | c.1009C>A | p.R337S |
| chr11 | 108170483 | 108170486 | TCTC | . | ATM | DEL | 0.06 | c.5047\_5051delTTCTC | p.F1683fs\*7 |
| chr11 | 108214019 | 108214019 | T | A | ATM | SNV | 0.05 | c.8339T>A | p.L2780H |
| chr11 | 125513686 | 125513686 | G | A | CHEK1 | SNV | 0.02 | c.815-1G>A | p.? |
| chr12 | 441055 | 441055 | C | T | KDM5A | SNV | 0.03 | c.1703G>A | p.R568H |
| chr12 | 6709009 | 6709009 | T | C | CHD4 | SNV | 0.02 | c.1412A>G | p.Y471C |
| chr12 | 25380276 | 25380276 | T | G | KRAS | SNV | 0.13 | c.182A>C | p.Q61P |
| chr12 | 121431498 | 121431498 | G | A | HNF1A | SNV | 0.13 | c.702G>A | p.E234E |
| chr13 | 28964091 | 28964091 | A | G | FLT1 | SNV | 0.03 | c.1811T>C | p.I604T |
| chr13 | 102375190 | 102375190 | C | A | FGF14 | SNV | 0.03 | c.750G>T | p.K250N |
| chr14 | 75514890 | 75514890 | G | T | MLH3 | SNV | 0.19 | c.1469C>A | p.S490Y |
| chr14 | 105246455 | 105246455 | C | T | AKT1 | SNV | 0.15 | c.145G>A | p.E49K |
| chr15 | 89811723 | 89811723 | T | C | FANCI | SNV | 0.08 | c.849T>C | p.Y283Y |
| chr15 | 90631838 | 90631838 | C | G | IDH2 | SNV | 0.07 | c.515G>C | p.R172T |
| chr15 | 93510648 | 93510648 | C | . | CHD2 | DEL | 0.05 | c.2094delC | p.R699fs\*35 |
| chr16 | 11348964 | 11348964 | C | T | SOCS1 | SNV | 0.03 | c.372G>A | p.T124T |
| chr16 | 67645347 | 67645348 | AA | . | CTCF | DEL | 0.03 | c.612\_613delAA | p.K205fs\*24 |
| chr16 | 89805927 | 89805927 | G | A | FANCA | SNV | 0.12 | c.3969C>T | p.A1323A |
| chr17 | 7577022 | 7577022 | G | A | TP53 | SNV | 0.06 | c.916C>T | p.R306\* |
| chr17 | 7578423 | 7578423 | C | A | TP53 | SNV | 0.07 | c.507G>T | p.M169I |
| chr17 | 11984671 | 11984671 | A | G | MAP2K4 | SNV | 0.06 | c.219-2A>G | p.? |
| chr17 | 37880997 | 37880997 | G | TTAT | ERBB2 | Sub\Complex\delins | 0.12 | c.2236delGinsTTAT | c.2326G>TTAT |
| chr17 | 37881440 | 37881440 | C | T | ERBB2 | SNV | 0.25 | c.2632C>T | p.H878Y |
| chr18 | 42530439 | 42530439 | A | C | SETBP1 | SNV | 0.05 | c.1134A>C | p.Q378H |
| chr18 | 45396908 | 45396908 | A | C | SMAD2 | SNV | 0.02 | c.264T>G | p.S88R |
| chr18 | 48603093 | 48603094 | . | T | SMAD4 | INS | 0.18 | c.1394\_1395insT | p.A466fs\*28 |
| 48603092 | 48603093 |
| chr19 | 36223270 | 36223270 | G | A | MLL4 | SNV | 0.08 | c.5826G>A | p.R1942R |
| chr2 | 25457251 | 25457251 | T | C | DNMT3A | SNV | 0.04 | c.2636A>G | p.N879S |
| chr2 | 25468160 | 25468160 | G | . | DNMT3A | DEL | 0.05 | c.1516\_1516delC | p.H506fs\*145 |
| chr2 | 29416770 | 29416770 | T | A | ALK | SNV | 0.03 | c.4183A>T | p.T1395S |
| chr2 | 29420411 | 29420411 | G | T | ALK | SNV | 0.16 | c.4070C>A | p.P1357H |
| chr2 | 29432664 | 29432664 | C | T | ALK | SNV | 0.2 | c.3824G>A | p.R1275Q |
| chr2 | 29443697 | 29443697 | A | G | ALK | SNV | 0.07 | c.3520T>C | p.F1174L |
| chr2 | 209113112 | 209113112 | C | A | IDH1 | SNV | 0.15 | c.395G>T | p.R132H |
| chr2 | 212251742 | 212251742 | C | A | ERBB4 | SNV | 0.05 | c.3317G>T | p.C1106F |
| chr22 | 30032779 | 30032800 | CGGACTCTGGGGCTCCGAGAAA | . | NF2 | DEL | 0.08 | c.154\_175del22 | p.R52fs\*64 |
| chr22 | 30050666 | 30050666 | T | A | NF2 | SNV | 0.12 | c.468T>A | p.S156R |
| chr3 | 10183532 | 10183548 | ATGCCCCGGAGGGCGGA | . | VHL | DEL | 0.12 | c.1\_17del17 | p.M1fs\*20 |
| 10183529 | 10183545 | GGAATGCCCCGGAGGGC |
| chr3 | 10183659 | 10183659 | C | T | VHL | SNV | 0.05 | c.128C>T | p.S43F |
| chr3 | 10188269 | 10188282 | CCATCTCTCAATGT | . | VHL | DEL | 0.03 | c.412\_425del14 | p.P138fs\*1 |
| chr3 | 10191626 | 10191630 | GCACA | . | VHL | DEL | 0.15 | c.619\_623delGCACA | p.A207fs\*>6 |
| chr3 | 25542806 | 25542806 | C | T | RARB | SNV | 0.03 | c.440C>T | p.S147F |
| chr3 | 41266107 | 41266107 | T | C | CTNNB1 | SNV | 0.08 | c.104T>C | p.I35T |
| chr3 | 47163586 | 47163587 | . | G | SETD2 | INS | 0.02 | c.1030\_1031insC | p.F344fs\*4 |
| chr3 | 52443861 | 52443861 | G | T | BAP1 | SNV | 0.25 | c.34C>A | p.P12T |
| chr3 | 142168302 | 142168302 | T | G | ATR | SNV | 0.07 | c.7904A>C | p.Q2635P |
| chr3 | 185161374 | 185161374 | G | A | MAP3K13 | SNV | 0.04 | c.801G>A | p.M267I |
| chr3 | 187447543 | 187447543 | C | T | BCL6 | SNV | 0.02 | c.650G>A | p.R217Q |
| chr3 | 192125845 | 192125845 | G | A | FGF12 | SNV | 0.03 | c.168C>T | p.S56S |
| chr4 | 55133541 | 55133541 | T | A | PDGFRA | SNV | 0.17 | c.845T>A | p.V282E |
| chr4 | 55155218 | 55155218 | G | T | PDGFRA | SNV | 0.12 | c.2817G>T | p.K939N |
| chr4 | 66356211 | 66356211 | A | G | EPHA5 | SNV | 0.16 | c.1286T>C | p.V429A |
| chr4 | 106157446 | 106157446 | G | T | TET2 | SNV | 0.02 | c.2347G>T | p.E783\* |
| chr4 | 153245453 | 153245453 | G | C | FBXW7 | SNV | 0.09 | c.1738C>G | p.H580D |
| chr4 | 153249518 | 153249519 | . | TGTCCCAC | FBXW7 | INS | 0.04 | c.1259\_1260insGTGGGACA | p.H420fs\*13 |
| chr5 | 56177964 | 56177964 | G | A | MAP3K1 | SNV | 0.02 | c.2448G>A | p.M816I |
| chr5 | 67589147 | 67589147 | A | G | PIK3R1 | SNV | 0.22 | c.1135A>G | p.K379E |
| chr5 | 112173983 | 112173984 | CA | . | APC | DEL | 0.18 | c.2692\_2693delCA | p.H898fs\*13 |
| chr5 | 112174579 | 112174579 | G | A | APC | SNV | 0.17 | c.3288G>A | p.Q1096Q |
| chr5 | 112176028 | 112176029 | . | A | APC | INS | 0.05 | c.4737\_4738insA | p.I1580fs\*11 |
| chr5 | 149495442 | 149495442 | C | T | PDGFRB | SNV | 0.12 | c.3205G>A | p.E1069K |
| chr5 | 180045774 | 180045774 | T | C | FLT4 | SNV | 0.07 | c.2997A>G | p.Q999Q |
| chr6 | 106552922 | 106552922 | G | T | PRDM1 | SNV | 0.02 | c.779G>T | p.R260L |
| chr6 | 117679018 | 117679018 | A | G | ROS1 | SNV | 0.18 | c.3803T>C | p.I1268T |
| chr6 | 157522541 | 157522541 | G | A | ARID1B | SNV | 0.04 | c.4759G>A | p.E1587K |
| chr6 | 170844466 | 170844466 | C | G | PSMB1 | SNV | 0.08 | c.568G>C | p.E190Q |
| chr7 | 2954933 | 2954933 | G | A | CARD11 | SNV | 0.08 | c.2777C>T | p.P926L |
| chr7 | 26224981 | 26224981 | G | A | NFE2L3 | SNV | 0.05 | c.1663G>A | p.V555I |
| chr7 | 55219011 | 55219011 | C | T | EGFR | SNV | 0.13 | c.584C>T | p.P195L |
| chr7 | 55241708 | 55241708 | G | C | EGFR | SNV | 0.08 | c.2156G>C; | p.G719A |
| chr7 | 55242427 | 55242427 | C | T | EGFR | SNV | 0.18 | c.2197C>T | p.P733S |
| chr7 | 55249012 | 55249013 | . | GGT | EGFR | INS | 0.15 | c.2310\_2311insGGT | p.D770\_N771insG |
| chr7 | 55249071 | 55249071 | C | T | EGFR | SNV | 0.04 | c.2369C>T | p.T790M |
| chr7 | 55259514 | 55259515 | TG | GA | EGFR | Sub | 0.02 | c.2573\_2574TG>GA | p.L858R |
| chr7 | 55259527 | 55259527 | T | A | EGFR | SNV | 0.07 | c.2585T>A | p.L862Q |
| chr7 | 140453136 | 140453136 | A | T | BRAF | SNV | 0.08 | c.1799T>A | p.V600E |
| chr9 | 5065019 | 5065019 | G | C | JAK2 | SNV | 0.11 | c.1193G>C | p.S398T |
| chr9 | 21970934 | 21970934 | G | A | CDKN2A | SNV | 0.04 | c.424C>T | p.H142Y |
| chr9 | 21974728 | 21974728 | C | A | CDKN2A | SNV | 0.1 | c.99G>T | p.E33D |
| chr9 | 36882061 | 36882061 | G | T | PAX5 | SNV | 0.03 | c.952C>A | p.H318N |
| chr9 | 133748371 | 133748371 | C | T | ABL1 | SNV | 0.06 | c.1032C>T | p.A344A |
| chr9 | 133760367 | 133760367 | C | A | ABL1 | SNV | 0.11 | c.2690C>A | p.P897H |
| chr9 | 135777018 | 135777019 | . | T | TSC1 | INS | 0.04 | c.2459\_2460insA | p.V821fs\*5 |
| chr9 | 139390791 | 139390792 | . | CC | NOTCH1 | INS | 0.06 | c.7399\_7400insGG | p.S2467fs\*11 |

注：上表黄色标注的预期结果两种书写格式均为正确结果。

**注意：**以下3个位点位于内含子区，不做评价：

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Chrom** | **Start** | **End** | **Ref** | **Alt** | **Gene** | **Type** | **VAF** | **CHGVS** | **PHGVS** |
| chr1 | 93299095 | 93299095 | C | T | RPL5 | SNV | 0.03 | c.74-7C>T | p.? |
| chr9 | 35074359 | 35074359 | G | A | FANCG | SNV | 0.09 | c.1760+9C>T | p.? |
| chr7 | 116411885 | 116411895 | CTTTCTCTCTG | . | MET | DEL | 0.07 | c.2942\_3082del141 | p.L982\_D1028del |

**Illumina-B1701-CNV\_SV预期结果**

**1，SV：**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Chr** | **Breakpoint 1** | **Breakpoint 2** | **Type** | **VAF (%)** | **Gene1** | **Gene2** | **Annotation** |
| *Chr10* | 32311365 | 43609712 | Inversion | 15 | KIF5B | RET | KIF5B-RET Fusion |

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Chr** | **Gene1** | **Breakpoint1** | **Type** | **Chr** | **Gene2** | **Breakpoint2** | **VAF(%)** | **Annotation** |
| *Chr11* | FLI1 | 128675350 | Translocation | chr22 | EWSR1 | 29684016 | 25 | FLI1-EWSR1 Fusion |

注意：Breakpoint位点允许有左右10个碱基以内的移位。

**2，CNV:**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Chr** | **Gene** | **Region** | **Type** | **VAF** |
| chr13 | FLT3 | Exon1-24 | CNV loss | 1 copy |
| chr8 | MYC | Exon1-3 | CNV gain | 4 copy |

**Illumina-B1702-SNV-Indel预期结果**

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **B1702** | **Chr** | **Start** | **End** | **Ref** | **Alt** | **Gene** | **Type** | **CHGVS** | **PHGVS** | **VAF(%)** |
| chr7 | 55242465 | 55242479 | GGAATTAAGAGAAGC | - | EGFR | Deletion | c.2235\_2249del15 | p.E746\_A750delELREA | 12 |
| chr3 | 41266113 | 41266113 | C | T | CTNNB1 | SNV | c.110C>T | p.S37F | 25 |
| chr7 | 55259515 | 55259515 | T | G | EGFR | SNV | c.2573T>G | p.L858R | 15 |
| chr10 | 89653782 | 89653782 | A | G | PTEN | SNV | c.80A>G | p.Y27C | 10 |

**Illumina-B1703-SNV-Indel预期结果**

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **B1703** | **Chr** | **Start** | **End** | **Ref** | **Alt** | **Gene** | **Type** | **CHGVS** | **PHGVS** | **VAF(%)** |
| chr7 | 55249071 | 55249071 | C | T | EGFR | SNV | c.2369C>T | p.T790M | 15 |
| chr7 | 55259515 | 55259515 | T | G | EGFR | SNV | c.2573T>G | p.L858R | 10 |
| chr3 | 41266113 | 41266113 | C | T | CTNNB1 | SNV | c.110C>T | p.S37F | 20 |
| chr16 | 2108752 | 2108752 | T | C | TSC2 | SNV | c.853T>C | p.Y285H | 15 |
| chr10 | 89653782 | 89653782 | A | G | PTEN | SNV | c.80A>G | p.Y27C | 8 |

**Illumina\_B1704\_SNV\_Indel预期结果**

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **B1704** | **Chr** | **Start** | **End** | **Ref** | **Alt** | **Gene** | **Type** | **CHGVS** | **PHGVS** | **VAF(%)** |
| chr7 | 55249071 | 55249071 | C | T | EGFR | SNV | c.2369C>T | p.T790M | 15 |
| chr7 | 55259515 | 55259515 | T | G | EGFR | SNV | c.2573T>G | p.L858R | 8 |
| chr7 | 55249092 | 55249092 | G | C | EGFR | SNV | c.2390G>C | p.C797S | 10 |
| chr16 | 2108752 | 2108752 | T | C | TSC2 | SNV | c.853T>C | p.Y285H | 25 |