



Sample Information

Patient Name: 莊佳敏**Gender:** Female**ID No.:** T222376968**History No.:** 44623236**Age:** 41**Ordering Doctor:** DOC1878G 沈佳儀**Ordering REQ.:** 0ALXFRD**Signing in Date:** 2019/11/05**Path No.:** S108-98788**MP No.:** TM1901**Assay:** Oncomine Tumor Mutation Load Assay**Sample Type:** FFPE**Block No.:** S108-47840C**Percentage of tumor cells:** 80%**Note:**

Sample Cancer Type: Non-Small Cell Lung Cancer

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Report Highlights

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Clinically Significant Biomarkers

■ Indicated ■ Contraindicated

| Genomic Alteration | Relevant Therapies (In this cancer type) | Relevant Therapies (In other cancer type) | Clinical Trials |
|---|---|--|-----------------|
| <i>EGFR exon 20 insertion</i> epidermal growth factor receptor Tier: IA Allele Frequency: 76.08% | ■ osimertinib ■ gefitinib ² | None | 58 |
| <i>Tumor Mutational Burden</i> 4.17 Mut/Mb measured | ■ ipilimumab + nivolumab ■ nivolumab | None | 11 |

Sources included in relevant therapies: FDA1, NCCN, EMA2, ESMO

Prevalent cancer biomarkers without clinical significance based on included data sources

RECQL4 splice site mutation



Tier Criteria Met

| Genomic Alteration | Tier Classification for Non-Small Cell Lung Cancer |
|---|--|
| <i>EGFR</i> exon 20 insertion Tier: IA | IA: Biomarker predicts response or resistance to FDA or EMA approved therapies in this cancer type IA: Biomarker is included in NCCN or ESMO guidelines that predict response or resistance to therapies in this cancer type IIC: Biomarker is an inclusion criteria for clinical trials |

Reference: Li et al. *Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer: A Joint Consensus Recommendation of the Association for Molecular Pathology, American Society of Clinical Oncology, and College of American Pathologists.* J Mol Diagn. 2017 Jan;19(1):4-23.

Variant Details

| DNA Sequence Variants | | | | | | | | |
|-----------------------|----------------------|--------------------------|------------|----------------|------------------|----------------|-------------------------|----------|
| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
| EGFR | p.(M766_A767insAS V) | c.2308_2309insCCA GCGTGG | COSM12376 | chr7:55248998 | 76.08% | NM_005228.4 | nonframeshift Insertion | 1990 |
| RECQL4 | | | . | chr8:145738767 | 100.00% | NM_004260.3 | | 1956 |
| TNFRSF14 | p.(K17R) | c.50A>G | . | chr1:2488153 | 36.05% | NM_003820.3 | missense | 1373 |
| MTOR | p.(=) | c.4731G>A | . | chr1:11205058 | 100.00% | NM_004958.3 | synonymous | 1302 |
| MTOR | p.(=) | c.2997C>T | . | chr1:11288758 | 99.88% | NM_004958.3 | synonymous | 1708 |
| MTOR | p.(=) | c.1437T>C | . | chr1:11301714 | 100.00% | NM_004958.3 | synonymous | 1998 |
| SDHB | p.(=) | c.18C>A | . | chr1:17380497 | 99.40% | NM_003000.2 | synonymous | 2000 |
| PAX7 | p.(=) | c.279C>T | . | chr1:18960990 | 38.32% | NM_002584.2 | synonymous | 1746 |
| MYCL | p.(T392S) | c.1175C>G | . | chr1:40363054 | 99.77% | NM_001033082.2 | missense | 1327 |
| MPL | p.(L265F) | c.793C>T | . | chr1:43805737 | 57.99% | NM_005373.2 | missense | 1690 |
| TAL1 | p.(=) | c.933A>G | . | chr1:47685455 | 99.91% | NM_003189.5 | synonymous | 1074 |
| CDKN2C | p.(=) | c.342C>T | . | chr1:51439777 | 42.39% | NM_078626.2 | synonymous | 1517 |
| JAK1 | p.(=) | c.2199A>G | . | chr1:65310489 | 49.25% | NM_002227.3 | synonymous | 1998 |
| LOC646626 | | | . | chr1:85741921 | 100.00% | NR_045484.1 | | 1656 |
| LOC646626 | | | . | chr1:85741968 | 48.25% | NR_045484.1 | | 1745 |
| DPYD | p.(R29C) | c.85C>T | . | chr1:98348885 | 99.79% | NM_000110.3 | missense | 1418 |
| TRIM33 | p.(I840T) | c.2519T>C | . | chr1:114948281 | 49.82% | NM_015906.3 | missense | 1084 |
| PDE4DIP | p.(=) | c.6771G>A | . | chr1:144855782 | 8.20% | NM_001198834.3 | synonymous | 61 |
| PDE4DIP | p.(=) | c.6687T>C | . | chr1:144855866 | 4.90% | NM_001198834.3 | synonymous | 2000 |
| PDE4DIP | p.(D1910E) | c.5730C>A | . | chr1:144865850 | 47.60% | NM_001198834.3 | missense | 1998 |
| PDE4DIP | p.(R1867C) | c.5599C>T | . | chr1:144866643 | 40.51% | NM_001198834.3 | missense | 1997 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|----------|-------------------|-----------|------------|----------------|------------------|----------------|----------------|----------|
| PDE4DIP | p.(=) | c.5580A>G | . | chr1:144866662 | 84.93% | NM_001198834.3 | synonymous | 1998 |
| PDE4DIP | p.(=) | c.4848A>G | . | chr1:144874760 | 33.93% | NM_001198834.3 | synonymous | 1998 |
| PDE4DIP | p.(H1598R) | c.4793A>G | . | chr1:144874815 | 29.03% | NM_001198834.3 | missense | 627 |
| PDE4DIP | p.(=) | c.4701C>G | . | chr1:144875979 | 36.30% | NM_001198834.3 | synonymous | 2000 |
| PDE4DIP | p.(K1454E) | c.4360A>G | . | chr1:144879090 | 20.65% | NM_001198834.3 | missense | 2000 |
| PDE4DIP | p.(=) | c.3438T>C | . | chr1:144882581 | 47.50% | NM_001198834.3 | synonymous | 2000 |
| PDE4DIP | p.(A1066T) | c.3196G>A | . | chr1:144882823 | 43.48% | NM_001198834.3 | missense | 1757 |
| PDE4DIP | p.(F1013I) | c.3037T>A | . | chr1:144886197 | 42.74% | NM_001198834.3 | missense | 1996 |
| PDE4DIP | p.(=) | c.2232C>T | . | chr1:144909956 | 17.51% | NM_001198834.3 | synonymous | 1999 |
| PDE4DIP | p.(R681H) | c.2042G>A | . | chr1:144912233 | 45.05% | NM_001198834.3 | missense | 2000 |
| PDE4DIP | p.(S536T) | c.1607G>C | . | chr1:144916748 | 45.57% | NM_001198834.3 | missense | 1997 |
| PDE4DIP | p.(=) | c.1558T>C | . | chr1:144917546 | 47.02% | NM_001198834.3 | synonymous | 1999 |
| PDE4DIP | p.(E410V) | c.1229A>T | . | chr1:144918957 | 44.17% | NM_001198834.3 | missense | 1999 |
| PDE4DIP | p.(S275L) | c.824C>T | . | chr1:144922583 | 43.70% | NM_001198834.3 | missense | 2000 |
| PDE4DIP | p.(R25L) | c.74G>T | . | chr1:144994658 | 44.91% | NM_001198834.3 | missense | 1995 |
| ITGA10 | p.(=) | c.1002T>C | . | chr1:145532549 | 99.14% | NM_003637.4 | synonymous | 1863 |
| ARNT | p.(=) | c.567G>C | . | chr1:150808889 | 73.29% | NM_001668.3 | synonymous | 1730 |
| DDR2 | p.(=) | c.1260C>G | . | chr1:162737116 | 71.56% | NM_006182.2 | synonymous | 1579 |
| PBX1 | p.(G21S) | c.61G>A | . | chr1:164529120 | 98.95% | NM_002585.3 | missense | 475 |
| RNASEL | p.(D541E) | c.1623T>G | . | chr1:182551337 | 33.01% | NM_021133.3 | missense | 1557 |
| MIR1278 | | | . | chr1:193104778 | 73.91% | NR_031691.1 | | 598 |
| PIK3C2B | p.(=) | c.2295C>T | . | chr1:204418364 | 26.29% | NM_002646.3 | synonymous | 1750 |
| PIK3C2B | p.(=) | c.1899C>T | . | chr1:204425028 | 67.59% | NM_002646.3 | synonymous | 108 |
| PIK3C2B | p.(=) | c.597G>A | . | chr1:204438334 | 74.84% | NM_002646.3 | synonymous | 1999 |
| PIK3C2B | p.(=) | c.288C>A | . | chr1:204438643 | 72.37% | NM_002646.3 | synonymous | 1998 |
| IKBKE | p.(=) | c.156A>G | . | chr1:206647742 | 99.45% | NM_014002.3 | synonymous | 1999 |
| MIR6769B | | | . | chr1:206647742 | 99.45% | NR_106919.1 | | 1999 |
| IKBKE | p.(=) | c.201T>C | . | chr1:206647787 | 99.56% | NM_014002.3 | synonymous | 1574 |
| MIR6769B | | | . | chr1:206647787 | 99.56% | NR_106919.1 | | 1574 |
| IKBKE | p.(=) | c.717G>A | . | chr1:206651107 | 99.00% | NM_014002.3 | synonymous | 1999 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|--------|-------------------|------------|------------|----------------|------------------|----------------|----------------|----------|
| IKBKE | p.(P713L) | c.2138C>T | . | chr1:206669465 | 69.83% | NM_014002.3 | missense | 1999 |
| PARP1 | p.(=) | c.1056A>G | . | chr1:226570840 | 96.48% | NM_001618.3 | synonymous | 1989 |
| MTR | p.(=) | c.3144A>G | . | chr1:237054569 | 75.60% | NM_000254.2 | synonymous | 2000 |
| MTR | p.(=) | c.3492C>A | . | chr1:237058744 | 68.99% | NM_000254.2 | synonymous | 1996 |
| MTR | p.(=) | c.3496C>T | . | chr1:237058748 | 29.20% | NM_000254.2 | synonymous | 2000 |
| MTR | p.(=) | c.3576C>T | . | chr1:237058828 | 72.74% | NM_000254.2 | synonymous | 1999 |
| FH | p.(P26L) | c.77C>T | . | chr1:241682946 | 24.35% | NM_000143.3 | missense | 1433 |
| NCOA1 | p.(=) | c.462G>C | . | chr2:24905927 | 100.00% | NM_003743.4 | synonymous | 1329 |
| NCOA1 | p.(=) | c.1512A>T | . | chr2:24929851 | 46.88% | NM_003743.4 | synonymous | 1538 |
| NCOA1 | p.(=) | c.3801T>G | . | chr2:24974945 | 99.90% | NM_003743.4 | synonymous | 1997 |
| ALK | p.(D1529E) | c.4587C>G | . | chr2:29416366 | 99.95% | NM_004304.4 | missense | 1998 |
| ALK | p.(K1491R) | c.4472A>G | . | chr2:29416481 | 99.60% | NM_004304.4 | missense | 1982 |
| ALK | p.(I1461V) | c.4381A>G | . | chr2:29416572 | 99.90% | NM_004304.4 | missense | 1999 |
| ALK | p.(=) | c.3375C>A | . | chr2:29445458 | 100.00% | NM_004304.4 | synonymous | 1997 |
| ALK | p.(=) | c.1500A>G | . | chr2:29543663 | 99.80% | NM_004304.4 | synonymous | 1995 |
| ALK | p.(=) | c.702T>A | . | chr2:29940529 | 96.60% | NM_004304.4 | synonymous | 589 |
| ALK | p.(=) | c.27C>G | . | chr2:30143499 | 99.80% | NM_004304.4 | synonymous | 1998 |
| EML4 | p.(K283E) | c.847A>G | . | chr2:42510018 | 99.12% | NM_019063.4 | missense | 1707 |
| EML4 | p.(I382V) | c.1144A>G | . | chr2:42515388 | 99.88% | NM_019063.4 | missense | 852 |
| MSH6 | p.(G39E) | c.116G>A | . | chr2:48010488 | 41.94% | NM_000179.2 | missense | 1545 |
| MSH6 | p.(=) | c.3306T>A | . | chr2:48030692 | 49.40% | NM_000179.2 | synonymous | 1994 |
| BCL11A | p.(=) | c.2088T>C | . | chr2:60687959 | 99.54% | NM_022893.3 | synonymous | 1961 |
| AFF3 | p.(S383N) | c.1148G>A | . | chr2:100343557 | 99.90% | NM_001025108.1 | missense | 1003 |
| LRP1B | p.(=) | c.13047G>A | . | chr2:141032088 | 99.58% | NM_018557.2 | synonymous | 959 |
| LRP1B | p.(=) | c.8526T>C | . | chr2:141260668 | 100.00% | NM_018557.2 | synonymous | 1021 |
| LRP1B | p.(=) | c.5256A>G | . | chr2:141571329 | 100.00% | NM_018557.2 | synonymous | 802 |
| LRP1B | p.(=) | c.2616C>T | . | chr2:141751592 | 58.25% | NM_018557.2 | synonymous | 1145 |
| SF3B1 | p.(=) | c.3657A>G | . | chr2:198257795 | 71.43% | NM_012433.3 | synonymous | 1995 |
| SF3B1 | p.(=) | c.2631T>C | . | chr2:198265526 | 67.15% | NM_012433.3 | synonymous | 1108 |
| SF3B1 | p.(=) | c.423A>G | . | chr2:198283305 | 71.07% | NM_012433.3 | synonymous | 1310 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|---------|-------------------|--------------------------|------------|----------------|------------------|----------------|----------------|----------|
| ERBB4 | p.(=) | c.3195A>G | . | chr2:212251864 | 99.94% | NM_005235.2 | synonymous | 1659 |
| FN1 | p.(V2261I) | c.6781G>A | . | chr2:216235089 | 100.00% | NM_212482.2 | missense | 1593 |
| FN1 | p.(=) | c.5691A>T | . | chr2:216242917 | 99.77% | NM_212482.2 | synonymous | 853 |
| FN1 | p.(=) | c.4725G>A | . | chr2:216249587 | 99.94% | NM_212482.2 | synonymous | 1730 |
| FN1 | p.(T817P) | c.2449A>C | . | chr2:216272900 | 100.00% | NM_212482.2 | missense | 1999 |
| FN1 | p.(Q15L) | c.44A>T | . | chr2:216300482 | 100.00% | NM_212482.2 | missense | 1305 |
| STK36 | p.(=) | c.2328G>A | . | chr2:219558698 | 15.83% | NM_015690.4 | synonymous | 1409 |
| STK36 | p.(=) | c.3237C>T | . | chr2:219563504 | 82.79% | NM_015690.4 | synonymous | 1778 |
| PAX3 | p.(?) | c.1451+3GCGTA>AA AGAC | . | chr2:223066128 | 6.57% | NM_181459.3 | unknown | 1980 |
| PAX3 | p.(=) | c.129T>C | . | chr2:223161889 | 99.92% | NM_181459.3 | synonymous | 1262 |
| CCDC140 | p.(?) | c.-6733A>G | . | chr2:223161889 | 99.92% | NM_153038.1 | unknown | 1262 |
| UGT1A8 | p.(P361L) | c.1082C>T | . | chr2:234676872 | 81.17% | NM_019076.4 | missense | 1689 |
| UGT1A10 | p.(P361L) | c.1082C>T | . | chr2:234676872 | 81.17% | NM_019075.2 | missense | 1689 |
| UGT1A9 | p.(P361L) | c.1082C>T | . | chr2:234676872 | 81.17% | NM_021027.2 | missense | 1689 |
| UGT1A7 | p.(P361L) | c.1082C>T | . | chr2:234676872 | 81.17% | NM_019077.2 | missense | 1689 |
| UGT1A6 | p.(P96L) | c.287C>T | . | chr2:234676872 | 81.17% | NM_205862.1 | missense | 1689 |
| UGT1A5 | p.(P365L) | c.1094C>T | . | chr2:234676872 | 81.17% | NM_019078.1 | missense | 1689 |
| UGT1A4 | p.(P365L) | c.1094C>T | . | chr2:234676872 | 81.17% | NM_007120.2 | missense | 1689 |
| UGT1A3 | p.(P365L) | c.1094C>T | . | chr2:234676872 | 81.17% | NM_019093.2 | missense | 1689 |
| UGT1A1 | p.(P364L) | c.1091C>T | . | chr2:234676872 | 81.17% | NM_000463.2 | missense | 1689 |
| CRBN | p.(=) | c.735T>C | . | chr3:3197918 | 26.04% | NM_016302.3 | synonymous | 937 |
| XPC | p.(Q939K) | c.2815C>A | . | chr3:14187449 | 100.00% | NM_004628.4 | missense | 1001 |
| XPC | p.(=) | c.1881T>A | . | chr3:14197987 | 100.00% | NM_004628.4 | synonymous | 1390 |
| XPC | p.(A499V) | c.1496C>T | . | chr3:14199887 | 23.86% | NM_004628.4 | missense | 1999 |
| XPC | p.(L16V) | c.46C>G | . | chr3:14220023 | 75.86% | NM_004628.4 | missense | 1628 |
| LSM3 | p.(?) | c.-338G>C | . | chr3:14220023 | 75.86% | NM_014463.2 | unknown | 1628 |
| TGFBR2 | p.(=) | c.1242C>T | . | chr3:30713842 | 25.16% | NM_001024847.2 | synonymous | 1228 |
| MLH1 | p.(R217C) | c.649C>T | . | chr3:37053562 | 20.27% | NM_000249.3 | missense | 587 |
| ITGA9 | p.(G507E) | c.1520G>A | . | chr3:37574951 | 99.73% | NM_002207.2 | missense | 1507 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|--------|-------------------|---------------|------------|----------------|------------------|----------------|-------------------------|----------|
| LTF | p.(=) | c.1894T>C | . | chr3:46480801 | 77.69% | NM_002343.5 | synonymous | 1067 |
| LTF | p.(E579D) | c.1737G>C | . | chr3:46480958 | 26.16% | NM_002343.5 | missense | 1827 |
| LTF | p.(=) | c.1623C>T | . | chr3:46484964 | 99.89% | NM_002343.5 | synonymous | 1744 |
| LTF | p.(K47R) | c.140A>G | . | chr3:46501213 | 99.37% | NM_002343.5 | missense | 1279 |
| LTF | p.(A29T) | c.85G>A | . | chr3:46501268 | 74.65% | NM_002343.5 | missense | 359 |
| LTF | p.(R23dup) | c.68_69insAAG | . | chr3:46501284 | 100.00% | NM_002343.5 | nonframeshift Insertion | 358 |
| SETD2 | p.(P1962L) | c.5885C>T | . | chr3:47125385 | 99.51% | NM_014159.6 | missense | 1017 |
| SETD2 | p.(=) | c.3465T>C | . | chr3:47162661 | 100.00% | NM_014159.6 | synonymous | 813 |
| PBRM1 | p.(=) | c.4335A>G | . | chr3:52584787 | 99.64% | NM_018313.4 | synonymous | 1374 |
| PBRM1 | p.(=) | c.3522A>T | . | chr3:52610651 | 99.62% | NM_018313.4 | synonymous | 1039 |
| PBRM1 | p.(=) | c.2211A>G | . | chr3:52643685 | 99.85% | NM_018313.4 | synonymous | 1334 |
| GATA2 | p.(=) | c.15C>G | . | chr3:128205860 | 99.70% | NM_032638.4 | synonymous | 1320 |
| EPHB1 | p.(=) | c.435C>T | . | chr3:134670524 | 34.07% | NM_004441.4 | synonymous | 1996 |
| EPHB1 | p.(=) | c.1800C>T | . | chr3:134898742 | 99.92% | NM_004441.4 | synonymous | 1229 |
| ATR | p.(=) | c.7875G>A | . | chr3:142168331 | 99.57% | NM_001184.3 | synonymous | 1395 |
| ATR | p.(=) | c.5208T>C | . | chr3:142222284 | 32.55% | NM_001184.3 | synonymous | 1155 |
| ATR | p.(=) | c.1815T>C | . | chr3:142277536 | 35.77% | NM_001184.3 | synonymous | 1339 |
| ATR | p.(=) | c.1776T>A | . | chr3:142277575 | 20.19% | NM_001184.3 | synonymous | 644 |
| ATR | p.(M211T) | c.632T>C | . | chr3:142281612 | 37.13% | NM_001184.3 | missense | 1080 |
| BCL6 | p.(=) | c.1161C>T | . | chr3:187447032 | 99.80% | NM_001706.4 | synonymous | 2000 |
| LPP | p.(=) | c.942C>T | . | chr3:188327461 | 66.37% | NM_005578.4 | synonymous | 1998 |
| TNK2 | p.(=) | c.273T>C | . | chr3:195615376 | 63.31% | NM_001010938.1 | synonymous | 1998 |
| FGFR3 | p.(=) | c.990C>T | . | chr4:1805478 | 98.34% | NM_000142.4 | synonymous | 963 |
| FGFR3 | p.(=) | c.1953G>A | . | chr4:1807894 | 99.91% | NM_000142.4 | synonymous | 1144 |
| PDGFRA | p.(=) | c.1701A>G | . | chr4:55141055 | 99.71% | NM_006206.5 | synonymous | 1732 |
| PDGFRA | p.(=) | c.3222T>C | . | chr4:55161391 | 99.93% | NM_006206.5 | synonymous | 1538 |
| KDR | p.(Q472H) | c.1416A>T | . | chr4:55972974 | 47.66% | NM_002253.2 | missense | 749 |
| ADGRL3 | p.(=) | c.612C>T | . | chr4:62598689 | 99.84% | NM_015236.5 | synonymous | 1287 |
| ADGRL3 | p.(=) | c.2079T>C | . | chr4:62800728 | 50.21% | NM_015236.5 | synonymous | 932 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|--------|-------------------|-----------|------------|----------------|------------------|----------------|----------------|----------|
| ADGRL3 | p.(=) | c.2709G>C | . | chr4:62845388 | 49.44% | NM_015236.5 | synonymous | 1962 |
| NFKB1 | p.(=) | c.1143T>C | . | chr4:103514658 | 99.80% | NM_003998.3 | synonymous | 1020 |
| TET2 | p.(I1762V) | c.5284A>G | . | chr4:106196951 | 99.94% | NM_001127208.2 | missense | 1743 |
| IL2 | p.(=) | c.114G>T | . | chr4:123377482 | 100.00% | NM_000586.3 | synonymous | 268 |
| SDHA | p.(Y55H) | c.163T>C | . | chr5:224487 | 49.65% | NM_004168.3 | missense | 1138 |
| SDHA | p.(=) | c.891T>C | . | chr5:231111 | 47.43% | NM_004168.3 | synonymous | 1849 |
| MTRR | p.(S284T) | c.850T>A | . | chr5:7878424 | 53.39% | NM_024010.2 | missense | 1208 |
| MTRR | p.(=) | c.1236G>A | . | chr5:7889216 | 54.17% | NM_024010.2 | synonymous | 1198 |
| MTRR | p.(R442C) | c.1324C>T | . | chr5:7889304 | 53.63% | NM_024010.2 | missense | 1613 |
| IL7R | p.(I66T) | c.197T>C | . | chr5:35861068 | 48.30% | NM_002185.4 | missense | 1236 |
| IL7R | p.(V138I) | c.412G>A | . | chr5:35871190 | 42.08% | NM_002185.4 | missense | 1925 |
| IL7R | p.(I356V) | c.1066A>G | . | chr5:35876274 | 48.65% | NM_002185.4 | missense | 2000 |
| IL7R | p.(D389Y) | c.1165G>T | . | chr5:35876373 | 26.59% | NM_002185.4 | missense | 1463 |
| APC | p.(=) | c.1458T>C | . | chr5:112162854 | 42.42% | NM_000038.5 | synonymous | 1589 |
| APC | p.(=) | c.4479G>A | . | chr5:112175770 | 99.20% | NM_000038.5 | synonymous | 1253 |
| APC | p.(=) | c.5034G>A | . | chr5:112176325 | 99.46% | NM_000038.5 | synonymous | 1114 |
| APC | p.(=) | c.5268T>G | . | chr5:112176559 | 99.47% | NM_000038.5 | synonymous | 937 |
| APC | p.(=) | c.5880G>A | . | chr5:112177171 | 100.00% | NM_000038.5 | synonymous | 735 |
| RAD50 | p.(?) | c.-38G>A | . | chr5:131892979 | 99.71% | NM_005732.3 | unknown | 1723 |
| CSF1R | p.(R748W) | c.2242C>T | . | chr5:149436927 | 74.57% | NM_005211.3 | missense | 1998 |
| CSF1R | p.(=) | c.726C>T | . | chr5:149457678 | 70.76% | NM_005211.3 | synonymous | 1997 |
| PDGFRB | p.(=) | c.3252A>G | . | chr5:149495395 | 99.93% | NM_002609.3 | synonymous | 1394 |
| PDGFRB | p.(=) | c.2601A>G | . | chr5:149499672 | 98.14% | NM_002609.3 | synonymous | 1989 |
| FGFR4 | p.(V10I) | c.28G>A | . | chr5:176516631 | 98.60% | NM_213647.2 | missense | 1999 |
| FGFR4 | p.(=) | c.162T>G | . | chr5:176517461 | 100.00% | NM_213647.2 | synonymous | 1998 |
| FGFR4 | p.(P136L) | c.407C>T | . | chr5:176517797 | 98.80% | NM_213647.2 | missense | 1997 |
| FGFR4 | p.(=) | c.702C>T | . | chr5:176518784 | 99.85% | NM_213647.2 | synonymous | 1995 |
| NSD1 | p.(=) | c.1482C>T | . | chr5:176636882 | 39.18% | NM_022455.4 | synonymous | 1654 |
| NSD1 | p.(=) | c.1749G>A | . | chr5:176637149 | 42.74% | NM_022455.4 | synonymous | 1996 |
| NSD1 | p.(S726P) | c.2176T>C | . | chr5:176637576 | 41.98% | NM_022455.4 | missense | 1584 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|--------|-------------------|------------------------|------------|----------------|------------------|----------------|---------------------------|----------|
| NSD1 | p.(=) | c.6829T>C | . | chr5:176721198 | 59.00% | NM_022455.4 | synonymous | 1995 |
| DEK | p.(=) | c.849G>A | . | chr6:18237661 | 100.00% | NM_003472.3 | synonymous | 655 |
| DEK | p.(=) | c.9C>T | . | chr6:18264210 | 99.36% | NM_003472.3 | synonymous | 939 |
| POU5F1 | p.(=) | c.63G>A | . | chr6:31138335 | 53.55% | NM_002701.5 | synonymous | 338 |
| NOTCH4 | p.(=) | c.2967A>C | . | chr6:32172065 | 100.00% | NM_004557.3 | synonymous | 1994 |
| NOTCH4 | p.(K117Q) | c.349A>C | . | chr6:32190390 | 60.70% | NM_004557.3 | missense | 2000 |
| NOTCH4 | p.(=) | c.333T>C | . | chr6:32190406 | 61.33% | NM_004557.3 | synonymous | 1999 |
| NOTCH4 | p.(L13_L16del) | c.36_47delGCTGCTGCTGCT | . | chr6:32191658 | 45.74% | NM_004557.3 | nonframeshift Deletion | |
| DAXX | p.(=) | c.1173T>C | . | chr6:33288271 | 62.64% | NM_001141970.1 | synonymous | 1997 |
| PKHD1 | p.(Q3899R) | c.11696A>G | . | chr6:51491884 | 48.75% | NM_138694.3 | missense | 1282 |
| PKHD1 | p.(=) | c.10521C>T | . | chr6:51524403 | 49.84% | NM_138694.3 | synonymous | 1603 |
| PKHD1 | p.(=) | c.9237G>A | . | chr6:51613177 | 99.69% | NM_138694.3 | synonymous | 1965 |
| PKHD1 | p.(=) | c.7764A>G | . | chr6:51720838 | 99.62% | NM_138694.3 | synonymous | 793 |
| PKHD1 | p.(L1870V) | c.5608T>G | . | chr6:51875250 | 99.85% | NM_138694.3 | missense | 1297 |
| PKHD1 | p.(V836A) | c.2507T>C | . | chr6:51910887 | 50.00% | NM_138694.3 | missense | 1882 |
| PKHD1 | p.(K626R) | c.1877A>G | . | chr6:51918923 | 50.45% | NM_138694.3 | missense | 1433 |
| PKHD1 | p.(Y617H) | c.1849T>C | . | chr6:51918951 | 49.02% | NM_138694.3 | missense | 1424 |
| PKHD1 | p.(=) | c.1185T>C | . | chr6:51924774 | 99.92% | NM_138694.3 | synonymous | 1248 |
| PKHD1 | p.(=) | c.234C>T | . | chr6:51947237 | 99.68% | NM_138694.3 | synonymous | 1546 |
| ICK | p.(=) | c.975A>G | . | chr6:52878637 | 56.64% | NM_014920.3 | synonymous | 761 |
| DST | p.(L4874V) | c.14620C>G | . | chr6:56351972 | 47.38% | NM_001144769.2 | missense | 1315 |
| DST | p.(R4599H) | c.13796G>A | . | chr6:56362247 | 47.31% | NM_001144769.2 | missense | 1655 |
| DST | p.(=) | c.10041G>A | . | chr6:56417192 | 50.59% | NM_001144769.2 | synonymous | 1194 |
| DST | p.(M3317I) | c.9951G>A | . | chr6:56417282 | 100.00% | NM_001144769.2 | missense | 1029 |
| DST | p.(T3230A) | c.9688A>G | . | chr6:56417545 | 51.68% | NM_001144769.2 | missense | 1341 |
| DST | p.(R2795H) | c.8384G>A | . | chr6:56420538 | 50.37% | NM_001144769.2 | missense | 1868 |
| DST | p.(=) | c.8184A>G | . | chr6:56422216 | 48.31% | NM_001144769.2 | synonymous | 1571 |
| ADGRB3 | p.(N503S) | c.1508A>G | . | chr6:69666684 | 99.77% | NM_001704.2 | missense | 436 |
| ADGRB3 | p.(=) | c.4008G>A | . | chr6:70071173 | 99.89% | NM_001704.2 | synonymous | 888 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|-------|-------------------|------------|------------|----------------|------------------|----------------|----------------|----------|
| EPHA7 | p.(=) | c.2076G>A | . | chr6:93967851 | 26.49% | NM_004440.3 | synonymous | 721 |
| EPHA7 | p.(=) | c.1341T>C | . | chr6:93982124 | 24.29% | NM_004440.3 | synonymous | 1128 |
| EPHA7 | p.(=) | c.981G>A | . | chr6:94067981 | 100.00% | NM_004440.3 | synonymous | 900 |
| PRDM1 | p.(G74S) | c.220G>A | . | chr6:106536253 | 21.36% | NM_001198.3 | missense | 1175 |
| ROS1 | p.(T145P) | c.433A>C | . | chr6:117725448 | 23.90% | NM_002944.2 | missense | 1071 |
| ROS1 | p.(=) | c.303A>T | . | chr6:117725578 | 100.00% | NM_002944.2 | synonymous | 1254 |
| SGK1 | p.(=) | c.633A>G | . | chr6:134494481 | 17.16% | NM_001143676.1 | synonymous | 1055 |
| ESR1 | p.(=) | c.30T>C | . | chr6:152129077 | 17.89% | NM_001122740.1 | synonymous | 1627 |
| ESR1 | p.(=) | c.729T>C | . | chr6:152201875 | 99.81% | NM_001122740.1 | synonymous | 1029 |
| ESR1 | p.(=) | c.975G>C | . | chr6:152265522 | 17.34% | NM_001122740.1 | synonymous | 1995 |
| ESR1 | p.(=) | c.1782G>A | . | chr6:152420095 | 16.80% | NM_001122740.1 | synonymous | 1762 |
| SYNE1 | p.(L8741M) | c.26221C>A | . | chr6:152443744 | 18.62% | NM_182961.3 | missense | 1676 |
| SYNE1 | p.(=) | c.25038T>C | . | chr6:152464839 | 99.51% | NM_182961.3 | synonymous | 1422 |
| SYNE1 | p.(F7302V) | c.21904T>G | . | chr6:152540278 | 97.99% | NM_182961.3 | missense | 1093 |
| SYNE1 | p.(T5426M) | c.16277C>T | . | chr6:152640110 | 16.93% | NM_182961.3 | missense | 1453 |
| SYNE1 | p.(=) | c.15597C>G | . | chr6:152646279 | 15.59% | NM_182961.3 | synonymous | 1649 |
| SYNE1 | p.(L5015M) | c.15043T>A | . | chr6:152647681 | 16.13% | NM_182961.3 | missense | 1147 |
| SYNE1 | p.(=) | c.14163C>T | . | chr6:152651657 | 82.78% | NM_182961.3 | synonymous | 1690 |
| SYNE1 | p.(S4596T) | c.13786T>A | . | chr6:152652034 | 17.29% | NM_182961.3 | missense | 827 |
| SYNE1 | p.(K4121R) | c.12362A>G | . | chr6:152658142 | 18.05% | NM_182961.3 | missense | 1014 |
| SYNE1 | p.(E4060D) | c.12180G>T | . | chr6:152665261 | 18.74% | NM_182961.3 | missense | 1724 |
| SYNE1 | p.(=) | c.10866T>C | . | chr6:152675854 | 19.41% | NM_182961.3 | synonymous | 1556 |
| SYNE1 | p.(=) | c.10191C>A | . | chr6:152683413 | 15.59% | NM_182961.3 | synonymous | 802 |
| SYNE1 | p.(=) | c.9495A>G | . | chr6:152694184 | 16.59% | NM_182961.3 | synonymous | 916 |
| SYNE1 | p.(V1035A) | c.3104T>C | . | chr6:152772264 | 100.00% | NM_182961.3 | missense | 150 |
| IGF2R | p.(L252V) | c.754C>G | . | chr6:160448324 | 82.30% | NM_000876.3 | missense | 1774 |
| IGF2R | p.(=) | c.1050A>G | . | chr6:160453978 | 100.00% | NM_000876.3 | synonymous | 432 |
| IGF2R | p.(=) | c.1590G>A | . | chr6:160464289 | 99.34% | NM_000876.3 | synonymous | 1956 |
| IGF2R | p.(=) | c.2139A>G | . | chr6:160468278 | 15.89% | NM_000876.3 | synonymous | 1429 |
| IGF2R | p.(R1619G) | c.4855A>G | . | chr6:160494409 | 16.45% | NM_000876.3 | missense | 1884 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|---------|-------------------|------------|------------|----------------|------------------|----------------|----------------|----------|
| IGF2R | p.(=) | c.6051C>G | . | chr6:160505199 | 100.00% | NM_000876.3 | synonymous | 1996 |
| RPS6KA2 | p.(=) | c.2221C>T | . | chr6:166826255 | 17.11% | NM_001006932.2 | synonymous | 1216 |
| RPS6KA2 | p.(=) | c.2172A>G | . | chr6:166826304 | 16.83% | NM_001006932.2 | synonymous | 1224 |
| RPS6KA2 | p.(=) | c.132C>T | . | chr6:166952264 | 99.76% | NM_001006932.2 | synonymous | 1681 |
| CARD11 | p.(=) | c.3276A>G | . | chr7:2946461 | 100.00% | NM_032415.5 | synonymous | 518 |
| CARD11 | p.(=) | c.2344C>T | . | chr7:2959172 | 52.13% | NM_032415.5 | synonymous | 1999 |
| CARD11 | p.(=) | c.2244G>C | . | chr7:2962293 | 63.98% | NM_032415.5 | synonymous | 1999 |
| PMS2 | p.(K541E) | c.1621A>G | . | chr7:6026775 | 99.25% | NM_000535.6 | missense | 1999 |
| PMS2 | p.(T485K) | c.1454C>A | . | chr7:6026942 | 52.60% | NM_000535.6 | missense | 1998 |
| PMS2 | p.(P470S) | c.1408C>T | . | chr7:6026988 | 46.65% | NM_000535.6 | missense | 2000 |
| PMS2 | p.(=) | c.780C>G | . | chr7:6036980 | 99.85% | NM_000535.6 | synonymous | 1996 |
| PMS2 | p.(=) | c.288C>T | . | chr7:6043386 | 54.19% | NM_000535.6 | synonymous | 1910 |
| ETV1 | p.(S100G) | c.298A>G | . | chr7:13978809 | 49.94% | NM_001163147.1 | missense | 847 |
| EGFR | p.(R521K) | c.1562G>A | . | chr7:55229255 | 98.80% | NM_005228.4 | missense | 1999 |
| EGFR | p.(=) | c.1887T>A | . | chr7:55238874 | 85.84% | NM_005228.4 | synonymous | 1992 |
| EGFR | p.(=) | c.2361G>A | . | chr7:55249063 | 87.63% | NM_005228.4 | synonymous | 1996 |
| EGFR | p.(=) | c.2709T>C | . | chr7:55266417 | 99.80% | NM_005228.4 | synonymous | 1982 |
| AKAP9 | p.(M463I) | c.1389G>T | . | chr7:91630620 | 28.28% | NM_005751.4 | missense | 1478 |
| AKAP9 | p.(=) | c.3075C>T | . | chr7:91632306 | 99.60% | NM_005751.4 | synonymous | 1513 |
| AKAP9 | p.(=) | c.3504A>G | . | chr7:91641928 | 55.43% | NM_005751.4 | synonymous | 1999 |
| AKAP9 | p.(N2792S) | c.8375A>G | . | chr7:91712698 | 55.74% | NM_005751.4 | missense | 1507 |
| AKAP9 | p.(=) | c.8665C>T | . | chr7:91713972 | 54.11% | NM_005751.4 | synonymous | 1508 |
| AKAP9 | p.(P2979S) | c.8935C>T | . | chr7:91714911 | 99.59% | NM_005751.4 | missense | 1234 |
| AKAP9 | p.(=) | c.9145C>T | . | chr7:91715662 | 60.56% | NM_005751.4 | synonymous | 1942 |
| AKAP9 | p.(=) | c.10426A>C | . | chr7:91726927 | 55.28% | NM_005751.4 | synonymous | 1402 |
| SAMD9 | p.(=) | c.1860A>G | . | chr7:92733551 | 47.18% | NM_001193307.1 | synonymous | 1509 |
| SAMD9 | p.(=) | c.960T>C | . | chr7:92734451 | 99.69% | NM_001193307.1 | synonymous | 1297 |
| EPHB4 | p.(=) | c.1752A>G | . | chr7:100411278 | 99.70% | NM_004444.4 | synonymous | 1995 |
| EPHB4 | p.(=) | c.1314T>C | . | chr7:100416250 | 99.90% | NM_004444.4 | synonymous | 2000 |
| PIK3CG | p.(=) | c.972A>G | . | chr7:106508978 | 99.75% | NM_002649.3 | synonymous | 1999 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|--------|-------------------|-----------|------------|----------------|------------------|----------------|----------------|----------|
| PIK3CG | p.(=) | c.981T>C | . | chr7:106508987 | 99.90% | NM_002649.3 | synonymous | 1999 |
| PIK3CG | p.(=) | c.2025C>T | . | chr7:106513011 | 49.73% | NM_002649.3 | synonymous | 1878 |
| MET | p.(=) | c.3912C>T | . | chr7:116435768 | 50.55% | NM_001127500.2 | synonymous | 2000 |
| MET | p.(=) | c.4071G>A | . | chr7:116436022 | 26.15% | NM_001127500.2 | synonymous | 1893 |
| MET | p.(=) | c.4146G>A | . | chr7:116436097 | 50.73% | NM_001127500.2 | synonymous | 1999 |
| SMO | p.(=) | c.1164G>C | . | chr7:128846328 | 99.75% | NM_005631.4 | synonymous | 1994 |
| WRN | p.(=) | c.513C>T | . | chr8:30924557 | 99.87% | NM_000553.4 | synonymous | 793 |
| WRN | p.(=) | c.2361G>T | . | chr8:30973957 | 27.34% | NM_000553.4 | synonymous | 545 |
| WRN | p.(L1074F) | c.3222G>T | . | chr8:30999280 | 23.50% | NM_000553.4 | missense | 583 |
| WRN | p.(=) | c.4083C>T | . | chr8:31024638 | 73.51% | NM_000553.4 | synonymous | 653 |
| NCOA2 | p.(M1282I) | c.3846G>C | . | chr8:71039118 | 54.37% | NM_006540.3 | missense | 1865 |
| NBN | p.(=) | c.2016A>G | . | chr8:90958422 | 35.40% | NM_002485.4 | synonymous | 2000 |
| NBN | p.(=) | c.1197T>C | . | chr8:90967711 | 99.73% | NM_002485.4 | synonymous | 1486 |
| NBN | p.(=) | c.102G>A | . | chr8:90995019 | 32.90% | NM_002485.4 | synonymous | 2000 |
| CSMD3 | p.(=) | c.6204T>C | . | chr8:113364696 | 35.55% | NM_198123.1 | synonymous | 1626 |
| CSMD3 | p.(=) | c.2121T>C | . | chr8:113702131 | 36.71% | NM_198123.1 | synonymous | 1204 |
| CSMD3 | p.(I219M) | c.657A>G | . | chr8:114186003 | 66.92% | NM_198123.1 | missense | 1998 |
| JAK2 | p.(=) | c.2490G>A | . | chr9:5081780 | 23.39% | NM_004972.3 | synonymous | 667 |
| PTPRD | p.(E1639D) | c.4917G>T | . | chr9:8341723 | 28.55% | NM_002839.3 | missense | 774 |
| PTPRD | p.(=) | c.4254G>C | . | chr9:8389364 | 74.30% | NM_002839.3 | synonymous | 747 |
| PTPRD | p.(R995C) | c.2983C>T | . | chr9:8485834 | 26.15% | NM_002839.3 | missense | 1602 |
| PTPRD | p.(Q447E) | c.1339C>G | . | chr9:8518052 | 72.47% | NM_002839.3 | missense | 1006 |
| TAF1L | p.(G1659R) | c.4975G>C | . | chr9:32630603 | 41.59% | NM_153809.2 | missense | 1998 |
| TAF1L | p.(A1229T) | c.3685G>A | . | chr9:32631893 | 14.52% | NM_153809.2 | missense | 1997 |
| PAX5 | p.(=) | c.1110C>T | . | chr9:36840623 | 66.24% | NM_016734.2 | synonymous | 1783 |
| PTCH1 | p.(P1315L) | c.3944C>T | . | chr9:98209594 | 20.30% | NM_000264.3 | missense | 2000 |
| PTCH1 | p.(=) | c.1686C>T | . | chr9:98238358 | 75.00% | NM_000264.3 | synonymous | 1644 |
| XPA | p.(?) | c.-4A>G | . | chr9:100459578 | 18.76% | NM_000380.3 | unknown | 986 |
| ABL1 | p.(=) | c.3324A>G | . | chr9:133761001 | 100.00% | NM_005157.5 | synonymous | 1999 |
| NUP214 | p.(P574S) | c.1720C>T | . | chr9:134020092 | 99.10% | NM_005085.3 | missense | 1218 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|---------|-------------------|-------------|------------|-----------------|------------------|----------------|----------------|----------|
| TSC1 | p.(=) | c.1335A>G | . | chr9:135782221 | 66.24% | NM_000368.4 | synonymous | 1004 |
| TSC1 | p.(M322T) | c.965T>C | . | chr9:135786904 | 63.80% | NM_000368.4 | missense | 1232 |
| BRD3 | p.(=) | c.1284C>T | . | chr9:136907005 | 98.99% | NM_007371.3 | synonymous | 1589 |
| BRD3 | p.(=) | c.936A>G | . | chr9:136913355 | 99.85% | NM_007371.3 | synonymous | 1997 |
| NOTCH1 | p.(=) | c.6555C>T | . | chr9:139391636 | 99.74% | NM_017617.4 | synonymous | 1535 |
| NOTCH1 | p.(=) | c.5094C>T | . | chr9:139397707 | 99.78% | NM_017617.4 | synonymous | 1791 |
| NOTCH1 | p.(=) | c.2265T>C | . | chr9:139407932 | 100.00% | NM_017617.4 | synonymous | 1997 |
| NOTCH1 | p.(?) | c.1441+7C>T | . | chr9:139412197 | 100.00% | NM_017617.4 | unknown | 1617 |
| NOTCH1 | p.(=) | c.312T>C | . | chr9:139418260 | 99.65% | NM_017617.4 | synonymous | 1720 |
| GATA3 | p.(P236A) | c.706C>G | . | chr10:8100732 | 48.90% | NM_001002295.1 | missense | 1996 |
| RET | p.(=) | c.135A>G | . | chr10:43595968 | 52.83% | NM_020975.4 | synonymous | 1999 |
| RET | p.(=) | c.1296A>G | . | chr10:43606687 | 99.75% | NM_020975.4 | synonymous | 1999 |
| RET | p.(=) | c.2307G>T | . | chr10:43613843 | 50.85% | NM_020975.4 | synonymous | 1237 |
| NCOA4 | p.(F8V) | c.22T>G | . | chr10:51568378 | 99.58% | NM_001145260.1 | missense | 1430 |
| NCOA4 | p.(=) | c.717C>T | . | chr10:51582894 | 49.62% | NM_001145260.1 | synonymous | 1707 |
| NCOA4 | p.(Q629P) | c.1886A>C | . | chr10:51586410 | 54.20% | NM_001145260.1 | missense | 1107 |
| TET1 | p.(?) | c.-30C>T | . | chr10:70332066 | 50.38% | NM_030625.2 | unknown | 1320 |
| TET1 | p.(D162G) | c.485A>G | . | chr10:70332580 | 50.82% | NM_030625.2 | missense | 1285 |
| TET1 | p.(I1123M) | c.3369A>G | . | chr10:70405855 | 53.17% | NM_030625.2 | missense | 756 |
| KAT6B | p.(E1364D) | c.4092G>C | . | chr10:76788674 | 27.04% | NM_012330.3 | missense | 995 |
| FAS | p.(=) | c.642T>C | . | chr10:90771829 | 99.76% | NM_000043.5 | synonymous | 831 |
| CYP2C19 | p.(=) | c.99T>C | . | chr10:96522561 | 51.62% | NM_000769.2 | synonymous | 1515 |
| CYP2C19 | p.(W212*) | c.636G>A | . | chr10:96540410 | 45.56% | NM_000769.2 | nonsense | 1420 |
| CYP2C19 | p.(=) | c.681G>A | . | chr10:96541616 | 45.40% | NM_000769.2 | synonymous | 663 |
| CYP2C19 | p.(=) | c.990C>T | . | chr10:96602622 | 50.75% | NM_000769.2 | synonymous | 1734 |
| CYP2C19 | p.(=) | c.1251A>C | . | chr10:96609775 | 50.25% | NM_000769.2 | synonymous | 804 |
| BLNK | p.(=) | c.171T>C | . | chr10:97990583 | 45.26% | NM_013314.3 | synonymous | 897 |
| NFKB2 | p.(=) | c.1269A>G | . | chr10:104159196 | 99.83% | NM_001077494.3 | synonymous | 572 |
| NFKB2 | p.(=) | c.1821A>G | . | chr10:104160434 | 99.83% | NM_001077494.3 | synonymous | 1165 |
| FGFR2 | p.(=) | c.696A>G | . | chr10:123298158 | 100.00% | NM_000141.4 | synonymous | 1661 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|----------|-------------------|------------|------------|-----------------|------------------|----------------|----------------|----------|
| RRM1 | p.(=) | c.2232G>A | . | chr11:4159466 | 38.69% | NM_001033.4 | synonymous | 654 |
| FANCF | p.(=) | c.786A>G | . | chr11:22646571 | 45.06% | NM_022725.3 | synonymous | 1305 |
| FANCF | p.(?) | c.-10C>T | . | chr11:22647366 | 56.25% | NM_022725.3 | unknown | 1321 |
| DDB2 | p.(=) | c.378T>C | . | chr11:47238522 | 99.95% | NM_000107.2 | synonymous | 1999 |
| MEN1 | p.(T546A) | c.1636A>G | . | chr11:64572018 | 49.22% | NM_000244.3 | missense | 1999 |
| MEN1 | p.(=) | c.1314T>C | . | chr11:64572557 | 99.95% | NM_000244.3 | synonymous | 1999 |
| MEN1 | p.(=) | c.1269C>T | . | chr11:64572602 | 51.65% | NM_000244.3 | synonymous | 1998 |
| CCND1 | p.(=) | c.723G>A | . | chr11:69462910 | 53.35% | NM_053056.2 | synonymous | 1700 |
| NUMA1 | p.(=) | c.2337G>A | . | chr11:71726212 | 46.85% | NM_006185.3 | synonymous | 1998 |
| BIRC3 | p.(=) | c.1200G>A | . | chr11:102201848 | 51.16% | NM_182962.2 | synonymous | 258 |
| ATM | p.(N1983S) | c.5948A>G | . | chr11:108183167 | 100.00% | NM_000051.3 | missense | 761 |
| KMT2A | p.(P3032A) | c.9094C>G | . | chr11:118375701 | 34.82% | NM_001197104.1 | missense | 1999 |
| CHEK1 | p.(I471V) | c.1411A>G | . | chr11:125525195 | 99.90% | NM_001274.5 | missense | 1920 |
| ETS1 | p.(=) | c.1143A>G | . | chr11:128333503 | 99.86% | NM_001143820.1 | synonymous | 1437 |
| FLI1 | p.(=) | c.279C>T | . | chr11:128638061 | 51.95% | NM_002017.4 | synonymous | 2000 |
| FLI1 | p.(=) | c.630A>G | . | chr11:128651893 | 46.52% | NM_002017.4 | synonymous | 1610 |
| FLI1 | p.(=) | c.687T>A | . | chr11:128675292 | 52.60% | NM_002017.4 | synonymous | 886 |
| KRAS | p.(=) | c.483G>A | . | chr12:25368462 | 99.56% | NM_033360.3 | synonymous | 1124 |
| ADAMTS20 | p.(=) | c.1296T>C | . | chr12:43860526 | 99.69% | NM_025003.4 | synonymous | 321 |
| ARID2 | p.(Y1299C) | c.3896A>G | . | chr12:46245802 | 51.06% | NM_152641.3 | missense | 995 |
| KMT2D | p.(=) | c.13689C>T | . | chr12:49424534 | 48.21% | NM_003482.3 | synonymous | 809 |
| KMT2D | p.(=) | c.7479G>T | . | chr12:49434074 | 44.19% | NM_003482.3 | synonymous | 1247 |
| ATF1 | p.(=) | c.327C>T | . | chr12:51203371 | 50.79% | NM_005171.4 | synonymous | 1144 |
| DDIT3 | p.(=) | c.99C>T | . | chr12:57911160 | 47.99% | NM_001195055.1 | synonymous | 1567 |
| HNF1A | p.(S574G) | c.1720A>G | . | chr12:121437382 | 99.93% | NM_000545.6 | missense | 1414 |
| EP400 | p.(=) | c.4758A>G | . | chr12:132508389 | 52.88% | NM_015409.4 | synonymous | 1999 |
| EP400 | p.(A3094T) | c.9280G>A | . | chr12:132562126 | 51.06% | NM_015409.4 | missense | 1835 |
| FLT3 | p.(=) | c.288C>T | . | chr13:28636084 | 99.92% | NM_004119.2 | synonymous | 1295 |
| FLT1 | p.(=) | c.3639C>T | . | chr13:28883061 | 100.00% | NM_002019.4 | synonymous | 399 |
| FLT1 | p.(=) | c.2901G>A | . | chr13:28896979 | 24.95% | NM_002019.4 | synonymous | 1912 |



Variant Details (continued)

| DNA Sequence Variants (continued) | | | | | | | | |
|-----------------------------------|-------------------|-----------|------------|-----------------|------------------|----------------|----------------|----------|
| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
| FLT1 | p.(=) | c.1704G>A | . | chr13:28964198 | 26.27% | NM_002019.4 | synonymous | 491 |
| FLT1 | p.(?) | c.-59C>T | . | chr13:29069039 | 24.85% | NM_002019.4 | unknown | 837 |
| BIVM-ERCC5 | p.(=) | c.1500T>C | . | chr13:103504517 | 31.48% | NM_001204425.1 | synonymous | 702 |
| ERCC5 | p.(=) | c.138T>C | . | chr13:103504517 | 31.48% | NM_000123.3 | synonymous | 702 |
| BIVM-ERCC5 | p.(G1507R) | c.4519G>C | . | chr13:103527849 | 99.85% | NM_001204425.1 | missense | 679 |
| ERCC5 | p.(G1053R) | c.3157G>C | . | chr13:103527849 | 99.85% | NM_000123.3 | missense | 679 |
| BIVM-ERCC5 | p.(G1534R) | c.4600G>C | . | chr13:103527930 | 99.85% | NM_001204425.1 | missense | 686 |
| ERCC5 | p.(G1080R) | c.3238G>C | . | chr13:103527930 | 99.85% | NM_000123.3 | missense | 686 |
| BIVM-ERCC5 | p.(D1558H) | c.4672G>C | . | chr13:103528002 | 66.27% | NM_001204425.1 | missense | 759 |
| ERCC5 | p.(D1104H) | c.3310G>C | . | chr13:103528002 | 66.27% | NM_000123.3 | missense | 759 |
| IRS2 | p.(G1057D) | c.3170G>A | . | chr13:110435231 | 38.81% | NM_003749.2 | missense | 371 |
| IRS2 | p.(=) | c.3099A>G | . | chr13:110435302 | 64.13% | NM_003749.2 | synonymous | 446 |
| IRS2 | p.(=) | c.2673G>C | . | chr13:110435728 | 68.70% | NM_003749.2 | synonymous | 591 |
| IRS2 | p.(=) | c.2448T>C | . | chr13:110435953 | 99.94% | NM_003749.2 | synonymous | 1634 |
| IRS2 | p.(=) | c.2169C>T | . | chr13:110436232 | 100.00% | NM_003749.2 | synonymous | 52 |
| LAMP1 | p.(=) | c.556C>A | . | chr13:113965176 | 100.00% | NM_005561.3 | synonymous | 1126 |
| BCL2L2-PABPN1 | p.(=) | c.123G>A | . | chr14:23777099 | 99.94% | NM_001199864.1 | synonymous | 1706 |
| BCL2L2 | p.(=) | c.123G>A | . | chr14:23777099 | 99.94% | NM_001199839.1 | synonymous | 1706 |
| BCL2L2-PABPN1 | p.(Q133R) | c.398A>G | . | chr14:23777374 | 100.00% | NM_001199864.1 | missense | 2000 |
| BCL2L2 | p.(Q133R) | c.398A>G | . | chr14:23777374 | 100.00% | NM_001199839.1 | missense | 2000 |
| NIN | p.(=) | c.5637G>A | . | chr14:51204996 | 100.00% | NM_020921.3 | synonymous | 1442 |
| NIN | p.(G1320E) | c.3959G>A | . | chr14:51223789 | 58.22% | NM_020921.3 | missense | 1673 |
| NIN | p.(Q1125P) | c.3374A>C | . | chr14:51224374 | 63.28% | NM_020921.3 | missense | 1999 |
| NIN | p.(P1111A) | c.3331C>G | . | chr14:51224417 | 65.23% | NM_020921.3 | missense | 1714 |
| NIN | p.(=) | c.1128T>C | . | chr14:51237701 | 40.00% | NM_020921.3 | synonymous | 2000 |
| NIN | p.(=) | c.933G>C | . | chr14:51239067 | 99.85% | NM_020921.3 | synonymous | 1999 |
| HIF1A | p.(P582S) | c.1744C>T | . | chr14:62207557 | 62.60% | NM_001530.3 | missense | 1441 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|----------|-------------------|-----------|------------|-----------------|------------------|----------------|----------------|----------|
| TSHR | p.(E727D) | c.2181G>C | . | chr14:81610583 | 60.63% | NM_000369.2 | missense | 1999 |
| HSP90AA1 | p.(=) | c.1446C>T | . | chr14:102550803 | 100.00% | NM_001017963.2 | synonymous | 725 |
| HSP90AA1 | p.(=) | c.282C>T | . | chr14:102568296 | 99.44% | NM_001017963.2 | synonymous | 360 |
| HSP90AA1 | p.(M71L) | c.211A>T | . | chr14:102568367 | 100.00% | NM_001017963.2 | missense | 1692 |
| AKT1 | p.(=) | c.726G>A | . | chr14:105239894 | 33.62% | NM_001014431.1 | synonymous | 1047 |
| THBS1 | p.(=) | c.1290G>A | . | chr15:39879717 | 18.85% | NM_003246.3 | synonymous | 2000 |
| THBS1 | p.(=) | c.1410C>T | . | chr15:39880358 | 18.49% | NM_003246.3 | synonymous | 1314 |
| THBS1 | p.(T523A) | c.1567A>G | . | chr15:39880822 | 22.26% | NM_003246.3 | missense | 1999 |
| KNL1 | p.(R43T) | c.128G>C | . | chr15:40898643 | 19.41% | NM_144508.4 | missense | 608 |
| KNL1 | p.(A460S) | c.1378G>T | . | chr15:40913840 | 22.88% | NM_144508.4 | missense | 883 |
| KNL1 | p.(R910G) | c.2728A>G | . | chr15:40915190 | 22.62% | NM_144508.4 | missense | 526 |
| KNL1 | p.(K1259E) | c.3775A>G | . | chr15:40916237 | 23.04% | NM_144508.4 | missense | 881 |
| TGM7 | p.(=) | c.1992C>T | . | chr15:43568794 | 84.35% | NM_052955.2 | synonymous | 1796 |
| TGM7 | p.(=) | c.1764G>C | . | chr15:43571390 | 84.08% | NM_052955.2 | synonymous | 1998 |
| TGM7 | p.(V515L) | c.1543G>T | . | chr15:43571958 | 84.75% | NM_052955.2 | missense | 2000 |
| TGM7 | p.(=) | c.777T>C | . | chr15:43579566 | 83.91% | NM_052955.2 | synonymous | 1585 |
| TGM7 | p.(G241E) | c.722G>A | . | chr15:43579621 | 81.55% | NM_052955.2 | missense | 1740 |
| TCF12 | p.(=) | c.135A>G | . | chr15:57213283 | 85.17% | NM_207037.1 | synonymous | 1234 |
| PML | p.(L217V) | c.649C>G | . | chr15:74315215 | 16.56% | NM_033238.2 | missense | 1999 |
| NTRK3 | p.(=) | c.678C>T | . | chr15:88679785 | 100.00% | NM_001012338.2 | synonymous | 1999 |
| NTRK3 | p.(=) | c.573C>T | . | chr15:88680684 | 68.13% | NM_001012338.2 | synonymous | 1999 |
| IGF1R | p.(=) | c.2298C>T | . | chr15:99465473 | 74.76% | NM_000875.4 | synonymous | 1997 |
| IGF1R | p.(=) | c.3129G>A | . | chr15:99478225 | 71.55% | NM_000875.4 | synonymous | 2000 |
| TSC2 | p.(R1268H) | c.3803G>A | . | chr16:2131788 | 78.54% | NM_000548.4 | missense | 1421 |
| CREBBP | p.(=) | c.6711C>T | . | chr16:3778337 | 19.10% | NM_004380.2 | synonymous | 513 |
| CREBBP | p.(=) | c.3900C>A | . | chr16:3795292 | 21.89% | NM_004380.2 | synonymous | 1448 |
| ERCC4 | p.(?) | c.-30T>A | . | chr16:14013993 | 80.26% | NM_005236.2 | unknown | 1140 |
| MYH11 | p.(A1241T) | c.3721G>A | . | chr16:15820863 | 99.85% | NM_001040114.1 | missense | 1998 |
| MYH11 | p.(=) | c.2493C>T | . | chr16:15839034 | 99.75% | NM_001040114.1 | synonymous | 1997 |
| MYH11 | p.(=) | c.1764T>C | . | chr16:15850204 | 99.55% | NM_001040114.1 | synonymous | 1997 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|-------|-------------------|------------------------|------------|----------------|------------------|----------------|----------------|----------|
| MYH11 | p.(=) | c.417C>T | . | chr16:15917197 | 14.25% | NM_001040114.1 | synonymous | 2000 |
| MMP2 | p.(=) | c.750C>T | . | chr16:55519607 | 57.78% | NM_004530.5 | synonymous | 1999 |
| MMP2 | p.(=) | c.1149T>C | . | chr16:55523705 | 99.90% | NM_004530.5 | synonymous | 1999 |
| MMP2 | p.(=) | c.1380G>A | . | chr16:55527113 | 55.22% | NM_004530.5 | synonymous | 1724 |
| CDH11 | p.(S373A) | c.1117T>G | . | chr16:65016087 | 99.85% | NM_001797.3 | missense | 1994 |
| CDH11 | p.(M275I) | c.825G>A | . | chr16:65022234 | 99.68% | NM_001797.3 | missense | 1549 |
| CDH5 | p.(=) | c.384C>T | . | chr16:66420885 | 99.90% | NM_001795.4 | synonymous | 1989 |
| CDH5 | p.(I517T) | c.1550_1551delTCins CT | . | chr16:66432423 | 99.90% | NM_001795.4 | missense | 1975 |
| CDH1 | p.(=) | c.2076T>C | . | chr16:68857441 | 67.43% | NM_004360.4 | synonymous | 1977 |
| FANCA | p.(=) | c.3807G>C | . | chr16:89807233 | 15.30% | NM_000135.3 | synonymous | 1144 |
| FANCA | p.(=) | c.2901C>T | . | chr16:89825065 | 16.50% | NM_000135.3 | synonymous | 2000 |
| FANCA | p.(G809D) | c.2426G>A | . | chr16:89836323 | 99.15% | NM_000135.3 | missense | 1999 |
| FANCA | p.(G501S) | c.1501G>A | . | chr16:89849480 | 99.53% | NM_000135.3 | missense | 1715 |
| FANCA | p.(A412V) | c.1235C>T | . | chr16:89857935 | 25.93% | NM_000135.3 | missense | 1697 |
| FANCA | p.(=) | c.1143G>T | . | chr16:89858417 | 16.38% | NM_000135.3 | synonymous | 1996 |
| FANCA | p.(T266A) | c.796A>G | . | chr16:89866043 | 99.69% | NM_000135.3 | missense | 982 |
| FANCA | p.(=) | c.115A>C | . | chr16:89882359 | 82.21% | NM_000135.3 | synonymous | 1647 |
| NLRP1 | p.(=) | c.4389C>T | . | chr17:5418107 | 99.72% | NM_033004.3 | synonymous | 1432 |
| NLRP1 | p.(R1366C) | c.4096C>T | . | chr17:5418799 | 99.74% | NM_033004.3 | missense | 1543 |
| NLRP1 | p.(=) | c.3741C>T | . | chr17:5424886 | 98.62% | NM_033004.3 | synonymous | 1381 |
| NLRP1 | p.(M1184V) | c.3550A>G | . | chr17:5425077 | 99.74% | NM_033004.3 | missense | 1135 |
| NLRP1 | p.(=) | c.2934A>G | . | chr17:5440197 | 99.65% | NM_033004.3 | synonymous | 1991 |
| NLRP1 | p.(=) | c.114G>C | . | chr17:5487164 | 76.52% | NM_033004.3 | synonymous | 1516 |
| TP53 | p.(C277F) | c.830G>T | . | chr17:7577108 | 58.90% | NM_000546.5 | missense | 2000 |
| TP53 | p.(P72R) | c.215C>G | . | chr17:7579472 | 73.82% | NM_000546.5 | missense | 1975 |
| PER1 | p.(A962P) | c.2884G>C | . | chr17:8046772 | 16.57% | NM_002616.2 | missense | 1949 |
| PER1 | p.(=) | c.639A>C | . | chr17:8053085 | 18.63% | NM_002616.2 | synonymous | 1444 |
| AURKB | p.(M298T) | c.893T>C | . | chr17:8108331 | 20.69% | NM_004217.3 | missense | 1498 |
| AURKB | p.(=) | c.885C>T | . | chr17:8108339 | 20.37% | NM_004217.3 | synonymous | 1507 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|--------|-------------------|--------------|------------|----------------|------------------|----------------|----------------|----------|
| ITGB3 | p.(=) | c.882T>C | . | chr17:45364540 | 53.86% | NM_000212.2 | synonymous | 1530 |
| ITGB3 | p.(=) | c.1143A>C | . | chr17:45368337 | 44.85% | NM_000212.2 | synonymous | 1496 |
| ITGB3 | p.(=) | c.1143A>T | . | chr17:45368337 | 54.95% | NM_000212.2 | synonymous | 1496 |
| ITGB3 | p.(=) | c.1533A>G | . | chr17:45369777 | 46.20% | NM_000212.2 | synonymous | 2000 |
| ITGB3 | p.(=) | c.1545G>A | . | chr17:45369789 | 45.64% | NM_000212.2 | synonymous | 1994 |
| COL1A1 | p.(T1075A) | c.3223A>G | . | chr17:48265495 | 99.90% | NM_000088.3 | missense | 1996 |
| COL1A1 | p.(=) | c.2298T>C | . | chr17:48268223 | 99.94% | NM_000088.3 | synonymous | 1740 |
| COL1A1 | p.(G130C) | c.388G>T | . | chr17:48276670 | 38.20% | NM_000088.3 | missense | 1979 |
| HLF | p.(?) | c.-50G>GTTTC | . | chr17:53342796 | 16.41% | NM_002126.4 | unknown | |
| BRIP1 | p.(=) | c.2637A>G | . | chr17:59763465 | 53.24% | NM_032043.2 | synonymous | 958 |
| CD79B | p.(=) | c.369T>C | . | chr17:62007498 | 48.45% | NM_001039933.2 | synonymous | 1994 |
| SEPT9 | p.(M576V) | c.1726A>G | . | chr17:75494705 | 100.00% | NM_001113491.1 | missense | 1609 |
| BIRC5 | p.(?) | c.-31G>C | . | chr17:76210367 | 47.56% | NM_001012271.1 | unknown | 1659 |
| BIRC5 | p.(E152K) | c.454G>A | . | chr17:76219591 | 50.75% | NM_001012271.1 | missense | 2000 |
| RNF213 | p.(N1045D) | c.3133A>G | . | chr17:78298938 | 99.65% | NM_001256071.2 | missense | 2000 |
| RNF213 | p.(=) | c.10470G>A | . | chr17:78327358 | 45.51% | NM_001256071.2 | synonymous | 1903 |
| ZNF521 | p.(=) | c.3597G>A | . | chr18:22775185 | 47.01% | NM_015461.2 | synonymous | 502 |
| CDH2 | p.(=) | c.2091T>C | . | chr18:25565082 | 100.00% | NM_001792.4 | synonymous | 1034 |
| CDH2 | p.(=) | c.1431C>G | . | chr18:25570228 | 97.82% | NM_001792.4 | synonymous | 641 |
| CDH2 | p.(A118T) | c.352G>A | . | chr18:25593694 | 45.56% | NM_001792.4 | missense | 1475 |
| CDH2 | p.(=) | c.333C>G | . | chr18:25593713 | 45.55% | NM_001792.4 | synonymous | 1473 |
| MBD1 | p.(P401A) | c.1201C>G | . | chr18:47800179 | 49.48% | NM_001204136.1 | missense | 1334 |
| MBD1 | p.(=) | c.240G>A | . | chr18:47803354 | 51.04% | NM_001204136.1 | synonymous | 1542 |
| DCC | p.(F23L) | c.67T>C | . | chr18:49867224 | 99.84% | NM_005215.3 | missense | 1234 |
| DCC | p.(R201G) | c.601C>G | . | chr18:50432602 | 98.08% | NM_005215.3 | missense | 1984 |
| DCC | p.(=) | c.2724A>G | . | chr18:50923713 | 49.76% | NM_005215.3 | synonymous | 822 |
| DCC | p.(=) | c.3108T>C | . | chr18:50936994 | 99.90% | NM_005215.3 | synonymous | 1037 |
| CDH20 | p.(?) | c.-23T>C | . | chr18:59157764 | 99.43% | NM_031891.3 | unknown | 529 |
| CDH20 | p.(=) | c.369C>T | . | chr18:59166541 | 49.06% | NM_031891.3 | synonymous | 1800 |
| CDH20 | p.(=) | c.780C>G | . | chr18:59170304 | 100.00% | NM_031891.3 | synonymous | 631 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|---------|-------------------|-----------|------------|----------------|------------------|----------------|----------------|----------|
| TCF3 | p.(=) | c.1302A>G | . | chr19:1619339 | 99.91% | NM_001136139.3 | synonymous | 1095 |
| KEAP1 | p.(=) | c.1611C>T | . | chr19:10599965 | 62.12% | NM_203500.1 | synonymous | 1930 |
| KEAP1 | p.(=) | c.1413C>G | . | chr19:10600442 | 99.77% | NM_203500.1 | synonymous | 1738 |
| PIK3R2 | p.(S313P) | c.937T>C | . | chr19:18273047 | 99.90% | NM_005027.3 | missense | 1997 |
| PIK3R2 | p.(=) | c.1911T>C | . | chr19:18279638 | 100.00% | NM_005027.3 | synonymous | 1996 |
| CCNE1 | p.(Q180K) | c.538C>A | . | chr19:30311684 | 63.58% | NM_001238.3 | missense | 755 |
| CCNE1 | p.(=) | c.1215C>T | . | chr19:30314666 | 66.25% | NM_001238.3 | synonymous | 1997 |
| AXL | p.(N266D) | c.796A>G | . | chr19:41743861 | 99.45% | NM_021913.4 | missense | 1649 |
| MARK4 | p.(=) | c.1683T>C | . | chr19:45801018 | 100.00% | NM_001199867.1 | synonymous | 1998 |
| ERCC2 | p.(=) | c.468A>C | . | chr19:45868309 | 65.88% | NM_000400.3 | synonymous | 1826 |
| ERCC1 | p.(=) | c.354T>C | . | chr19:45923653 | 99.88% | NM_001983.3 | synonymous | 1673 |
| ASXL1 | p.(L815P) | c.2444T>C | . | chr20:31022959 | 99.66% | NM_015338.5 | missense | 1747 |
| ASXL1 | p.(=) | c.3759T>C | . | chr20:31024274 | 100.00% | NM_015338.5 | synonymous | 1819 |
| SRC | p.(=) | c.1218G>C | . | chr20:36030939 | 99.58% | NM_198291.2 | synonymous | 1918 |
| PLCG1 | p.(I813T) | c.2438T>C | . | chr20:39797465 | 57.51% | NM_002660.2 | missense | 1998 |
| MIR6871 | | | . | chr20:39797465 | 57.51% | NR_106931.1 | | 1998 |
| PTPRT | p.(=) | c.2904C>T | . | chr20:40757394 | 28.10% | NM_133170.3 | synonymous | 2000 |
| PTPRT | p.(E808G) | c.2423A>G | . | chr20:40828005 | 15.01% | NM_133170.3 | missense | 1999 |
| AURKA | p.(I57V) | c.169A>G | . | chr20:54961463 | 55.06% | NM_003600.3 | missense | 1998 |
| GNAS | p.(=) | c.393C>T | . | chr20:57478807 | 99.80% | NM_000516.5 | synonymous | 1999 |
| ERG | p.(?) | c.-6C>T | . | chr21:39870310 | 99.88% | NM_182918.3 | unknown | 844 |
| ITGB2 | p.(=) | c.1323T>C | . | chr21:46311813 | 99.80% | NM_000211.4 | synonymous | 1999 |
| ITGB2 | p.(Q354H) | c.1062A>T | . | chr21:46314907 | 99.58% | NM_000211.4 | missense | 1660 |
| TIMP3 | p.(=) | c.249T>C | . | chr22:33253280 | 56.88% | NM_000362.4 | synonymous | 1591 |
| MYH9 | p.(I1626V) | c.4876A>G | . | chr22:36684354 | 99.75% | NM_002473.5 | missense | 1611 |
| MYH9 | p.(=) | c.4872G>T | . | chr22:36684358 | 99.75% | NM_002473.5 | synonymous | 1613 |
| MYH9 | p.(=) | c.3429T>G | . | chr22:36691607 | 99.93% | NM_002473.5 | synonymous | 1381 |
| EP300 | p.(=) | c.3183T>A | . | chr22:41551039 | 98.48% | NM_001429.3 | synonymous | 985 |
| CYP2D6 | p.(C296R) | c.886T>C | . | chr22:42523943 | 99.85% | NM_000106.5 | missense | 1998 |
| CYP2D6 | p.(P34S) | c.100C>T | . | chr22:42526694 | 99.10% | NM_000106.5 | missense | 2000 |



Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Variant ID | Locus | Allele Frequency | Transcript | Variant Effect | Coverage |
|--------|-------------------|-----------|------------|----------------|------------------|-------------|----------------|----------|
| KDM6A | p.(T726K) | c.2177C>A | . | chrX:44929077 | 100.00% | NM_021140.3 | missense | 1368 |
| TAF1 | p.(R1193Q) | c.3578G>A | . | chrX:70617214 | 48.71% | NM_004606.4 | missense | 657 |
| ATRX | p.(Q929E) | c.2785C>G | . | chrX:76937963 | 36.79% | NM_000489.4 | missense | 723 |
| MAGEA1 | p.(=) | c.813C>T | . | chrX:152482198 | 20.00% | NM_004988.4 | synonymous | 1640 |
| MAGEA1 | p.(T32A) | c.94A>G | . | chrX:152482917 | 18.74% | NM_004988.4 | missense | 1473 |

Relevant Therapy Summary

● In this cancer type
 ○ In other cancer type
 ⓘ In this cancer type and other cancer types
 ⛔ Contraindicated
 ⚠ Both for use and contraindicated
 ✕ No evidence

EGFR exon 20 insertion

| Relevant Therapy | FDA | NCCN | EMA | ESMO | Clinical Trials* |
|--|-----|------|-----|------|------------------|
| osimertinib | ✕ | ● | ✕ | ✕ | ● (II) |
| gefitinib | ✕ | ✕ | ⛔ | ✕ | ● (III) |
| apatinib + erlotinib, apatinib + gefitinib, apatinib + icotinib hydrochloride | ✕ | ✕ | ✕ | ✕ | ● (IV) |
| apatinib + gefitinib | ✕ | ✕ | ✕ | ✕ | ● (IV) |
| erlotinib + natural product, erlotinib + placebo, gefitinib + natural product, gefitinib + placebo, icotinib hydrochloride + natural product, icotinib hydrochloride + placebo | ✕ | ✕ | ✕ | ✕ | ● (IV) |
| gefitinib, radiation therapy | ✕ | ✕ | ✕ | ✕ | ● (IV) |
| icotinib hydrochloride, radiation therapy | ✕ | ✕ | ✕ | ✕ | ● (IV) |
| atezolizumab, bevacizumab, chemotherapy | ✕ | ✕ | ✕ | ✕ | ● (III) |
| bevacizumab + chemotherapy, bevacizumab (Shanghai Hengrui Pharmaceutical) + chemotherapy | ✕ | ✕ | ✕ | ✕ | ● (III) |
| chemotherapy, nivolumab | ✕ | ✕ | ✕ | ✕ | ● (III) |
| erlotinib, gefitinib | ✕ | ✕ | ✕ | ✕ | ● (III) |
| afatinib + bevacizumab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| afatinib + chemotherapy + radiation therapy + surgical intervention | ✕ | ✕ | ✕ | ✕ | ● (II) |

* Most advanced phase (IV, III, II/III, II, I/II, I) is shown and multiple clinical trials may be available.



Relevant Therapy Summary (continued)

● In this cancer type ○ In other cancer type ● In this cancer type and other cancer types ⚡ Contraindicated ⚠ Both for use and contraindicated ✕ No evidence

EGFR exon 20 insertion (continued)

| Relevant Therapy | FDA | NCCN | EMA | ESMO | Clinical Trials* |
|---|-----|------|-----|------|------------------|
| anlotinib hydrochloride + sintilimab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| apatinib + chemotherapy | ✕ | ✕ | ✕ | ✕ | ● (II) |
| bevacizumab, osimertinib | ✕ | ✕ | ✕ | ✕ | ● (II) |
| chemotherapy, ramucirumab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| erlotinib | ✕ | ✕ | ✕ | ✕ | ● (II) |
| erlotinib + chemotherapy | ✕ | ✕ | ✕ | ✕ | ● (II) |
| erlotinib + radiation therapy | ✕ | ✕ | ✕ | ✕ | ● (II) |
| gefitinib + chemotherapy | ✕ | ✕ | ✕ | ✕ | ● (II) |
| icotinib hydrochloride | ✕ | ✕ | ✕ | ✕ | ● (II) |
| ipilimumab, nivolumab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| KN046 | ✕ | ✕ | ✕ | ✕ | ● (II) |
| poziotinib | ✕ | ✕ | ✕ | ✕ | ● (II) |
| radiation therapy, tyrosine kinase inhibitors | ✕ | ✕ | ✕ | ✕ | ● (II) |
| sintilimab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| sunitinib | ✕ | ✕ | ✕ | ✕ | ● (II) |
| targeted therapy, targeted therapy + chemotherapy | ✕ | ✕ | ✕ | ✕ | ● (II) |
| tarloxotinib | ✕ | ✕ | ✕ | ✕ | ● (II) |
| afatinib + necitumumab | ✕ | ✕ | ✕ | ✕ | ● (I/II) |
| bevacizumab + erlotinib + chemotherapy | ✕ | ✕ | ✕ | ✕ | ● (I/II) |
| cetuximab, cetuximab + natural killer cell therapy | ✕ | ✕ | ✕ | ✕ | ● (I/II) |
| EMB01 | ✕ | ✕ | ✕ | ✕ | ● (I/II) |
| gefitinib + ningetinib | ✕ | ✕ | ✕ | ✕ | ● (I/II) |
| icotinib hydrochloride + chemotherapy + radiation therapy | ✕ | ✕ | ✕ | ✕ | ● (I/II) |
| oleclumab + osimertinib | ✕ | ✕ | ✕ | ✕ | ● (I/II) |
| TAK788 | ✕ | ✕ | ✕ | ✕ | ● (I/II) |

* Most advanced phase (IV, III, II/III, II, I/II, I) is shown and multiple clinical trials may be available.



Relevant Therapy Summary (continued)

● In this cancer type ○ In other cancer type ● In this cancer type and other cancer types ⛔ Contraindicated ⚠ Both for use and contraindicated ✕ No evidence

EGFR exon 20 insertion (continued)

| Relevant Therapy | FDA | NCCN | EMA | ESMO | Clinical Trials* |
|---|-----|------|-----|------|------------------|
| cetuximab + FATE-NK100 | ✕ | ✕ | ✕ | ✕ | ● (I) |
| durvalumab + oleclumab, oleclumab | ✕ | ✕ | ✕ | ✕ | ● (I) |
| erlotinib + ixazomib | ✕ | ✕ | ✕ | ✕ | ● (I) |
| everolimus + neratinib, neratinib + palbociclib, neratinib + trametinib | ✕ | ✕ | ✕ | ✕ | ● (I) |
| JNJ-61186372 | ✕ | ✕ | ✕ | ✕ | ● (I) |
| necitumumab, osimertinib | ✕ | ✕ | ✕ | ✕ | ● (I) |
| osimertinib, osimertinib + radiation therapy | ✕ | ✕ | ✕ | ✕ | ● (I) |
| pirotinib | ✕ | ✕ | ✕ | ✕ | ● (I) |
| TP-0903 | ✕ | ✕ | ✕ | ✕ | ● (I) |
| tyrosine kinase inhibitors, tyrosine kinase inhibitors + chemotherapy | ✕ | ✕ | ✕ | ✕ | ● (I) |

Tumor Mutational Burden

| Relevant Therapy | FDA | NCCN | EMA | ESMO | Clinical Trials* |
|---|-----|------|-----|------|------------------|
| ipilimumab + nivolumab | ✕ | ● | ✕ | ● | ✕ |
| nivolumab | ✕ | ● | ✕ | ✕ | ✕ |
| atezolizumab | ✕ | ✕ | ✕ | ✕ | ● (II/III) |
| durvalumab + tremelimumab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| ipilimumab + nivolumab, nivolumab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| ipilimumab + nivolumab, pembrolizumab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| ipilimumab, nivolumab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| lenvatinib, MK-1308, MK-4280, pembrolizumab | ✕ | ✕ | ✕ | ✕ | ● (II) |
| pembrolizumab, pembrolizumab + chemotherapy | ✕ | ✕ | ✕ | ✕ | ● (II) |
| sintilimab | ✕ | ✕ | ✕ | ✕ | ● (I/II) |
| KN046 | ✕ | ✕ | ✕ | ✕ | ● (I) |

* Most advanced phase (IV, III, II/III, II, I/II, I) is shown and multiple clinical trials may be available.



Relevant Therapy Details

Current NCCN Information

☒ In this cancer type
 ☐ In other cancer type
 ☐ In this cancer type and other cancer types
 ☐ Contraindicated
 ☐ Not recommended
 ☐ Resistance

NCCN information is current as of 2019-05-15. For the most up-to-date information, search www.nccn.org.
 For NCCN International Adaptations & Translations, search www.nccn.org/global/international_adaptations.aspx.

EGFR exon 20 insertion

● osimertinib

Cancer type: Non-Small Cell Lung Cancer

Variant class: EGFR mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- Non-Small Cell Lung Cancer; Brain metastases; Newly diagnosed (Not specified)
- Non-Small Cell Lung Cancer; Leptomeningeal and Spine metastases (Not specified)

Reference: NCCN Guidelines® - NCCN-Central Nervous System Cancers [Version 1.2019]

🗨️ pembrolizumab

Cancer type: Non-Small Cell Lung Cancer

Variant class: EGFR mutation

Other criteria: CD274 overexpression

Summary:

NCCN Guidelines® include the following supporting statement(s):

- "A small study suggests that single-agent pembrolizumab is not effective as first-line therapy in patients with metastatic NSCLC and EGFR mutations, even those with PD-L1 levels more than 50%."

Reference: NCCN Guidelines® - NCCN-Non-Small Cell Lung Cancer [Version 5.2019]

🛡️ EGFR tyrosine kinase inhibitor

Cancer type: Non-Small Cell Lung Cancer

Variant class: EGFR exon 20 insertion

Summary:

NCCN Guidelines® include the following supporting statement(s):

- "Patients with EGFR exon 20 insertion mutations are usually resistant to TKIs, although there are rare exceptions."

Reference: NCCN Guidelines® - NCCN-Non-Small Cell Lung Cancer [Version 5.2019]



Tumor Mutational Burden

● ipilimumab + nivolumab

Cancer type: Non-Small Cell Lung Cancer

Variant class: Tumor Mutational Burden

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- Non-Small Cell Lung Cancer; Emerging targeted agents

Reference: NCCN Guidelines® - NCCN-Non-Small Cell Lung Cancer [Version 5.2019]

● nivolumab

Cancer type: Non-Small Cell Lung Cancer

Variant class: Tumor Mutational Burden

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- Non-Small Cell Lung Cancer; Emerging targeted agents

Reference: NCCN Guidelines® - NCCN-Non-Small Cell Lung Cancer [Version 5.2019]



Current EMA Information

☒ In this cancer type ☐ In other cancer type ☐ In this cancer type and other cancer types ☒ Contraindicated ☒ Not recommended ☒ Resistance

EMA information is current as of 2019-08-23. For the most up-to-date information, search www.ema.europa.eu/ema.

EGFR exon 20 insertion

☒ gefitinib

Cancer type: Non-Small Cell Lung Cancer

Label as of: 2019-05-28

Variant class: EGFR exon 20 insertion

Reference:

https://www.ema.europa.eu/en/documents/product-information/iressa-epar-product-information_en.pdf



Current ESMO Information

☒ In this cancer type ☐ In other cancer type ☐ In this cancer type and other cancer types ☒ Contraindicated ☒ Not recommended ☒ Resistance

ESMO information is current as of 2019-05-15. For the most up-to-date information, search www.esmo.org.

Tumor Mutational Burden

☒ ipilimumab + nivolumab

Cancer type: Non-Small Cell Lung Cancer

Variant class: Tumor Mutational Burden

ESMO Level of Evidence/Grade of Recommendation: I / A

Population segment (Line of therapy):

- Stage IV Squamous and Non-squamous Non-Small Cell Lung Cancer (First-line therapy)

Reference: ESMO Clinical Practice Guidelines - ESMO-Metastatic Non-Small-Cell Lung Cancer [Ann Oncol (2018) 29 (suppl 4): iv192–iv237. (Corrigendum: 30 January 2019)]

Signatures

Testing Personnel:

Laboratory Supervisor:

Pathologist: