| Tests by gene, mutation type and codon(s) tested: | | |
|---|---|---|
| Gene | Mutation Type | Codon(s) Tested |
| ABL1 | single base substitution | 244, 248, 250, 252, 253, 255, 276, 315, 317, 351, 355, 359, 384, 387, 396 |
| AKT1 | single base substitution | 17, 35, 49, 173, 179 |
| ALK | single base substitution | 1174, 1196, 1275 |
| APC | single base substitution | 876, 880, 886, 1096, 1114, 1120, 1291, 1294, 1295, 1300, 1301, 1303, 1306, 1308, 1309, 1310, 1312, 1315, 1317, 1319, 1322, 1345, 1346, 1350, 1353, 1355, 1356, 1366, 1367, 1370, 1374, 1376, 1378, 1379, 1429, 1432, 1435, 1442, 1443, 1444, 1447, 1450, 1451, 1454, 1459, 1461, 1464, 1469, |
| APC | insertion/deletion | 869, 1294, 1300, 1301, 1303, 1307, 1309, 1310, 1311, 1312, 1314, 1315, 1319, 1348, 1351, 1355, 1356, 1357, 1361, 1364, 1372, 1373, 1375, 1376, 1378, 1431, 1435, 1436, 1438, 1439, 1440, 1443, 1445, 1449, 1453, 1455, 1458, 1459, 1462, 1464, 1465, 1488, 1489, 1490, 1491, 1492, 1493, 1494, |
| AR | single base substitution | 45,46,56,57,58,59,66,67,75,264,269,288,289,290,355,364,547,548,553,565,568,573,638,640,654,663,671,675,828,830,832,839,840,848,856,857,86 |
| AR | insertion/deletion | 79,551, |
| ATM | single base substitution | 337, 410, 604, 848, 858, 1309, 1681, 1682, 1742, 1794, 1941, 2442, 2443, 2666, 2695, 2725, 2888, 2890, 2947, 3008, 3047 |
| ATM | insertion/deletion | 353 |
| ATR | single base substitution | 2587,2580,2579,2575,2549,2540,2537,2533,2438,2437,2431,2428,2425,2419,2409,2407,2402,1849,1831,1724,1716,1707,1699,1095,1082,1081,1078,1061,766,757,706,704,700,699,695,91,76,73,65, |
| ATR | insertion/deletion | 774, |
| AURKA | single base substitution | 86,81, |
| AURKB | single base substitution | 128,126, |
| AXL | single base substitution | 200,273,285,289,293,295,304,742,744,745,747, |
| AXL | insertion/deletion | 292, |
| BARD1 | single base substitution | 92,89,85,24,21, |
| BLM | single base substitution | 229,233,241,565,589,600,1127,1132,1140,1142,1144,1279,1280,1283,132 |
| BLM | insertion/deletion | 254,1308, |
| BRAF | single base substitution | 444, 453, 462, 464, 466, 469, 471, 581, 582, 583, 584, 586, 587, 592, 594, 595, 596, 597, 598, 599, 600, 601, 603, 604, 605, 606, 608 |
| BRAF | insertion/deletion | 601, 598-600, 604 |

| | | |
|---|---|---|
| BRCA1 | single base substitution | 1819,1816,1811,1794,1788,1751,1749,1350,1344,1342,1335,1309,1301,1281,1276,1140,1123,1115,1104,1040,1033,1027,1020,1014,1011,1009,996,988,986,942,935,934,907,793,778,758,694,690,687,673,644,628,461,453,451,448,446,431,374,372,370,362,321,320,309,297,275,272,57,38,30,9, |
| BRCA1 | insertion/deletion | 1814,1796,1756,1749,1309,1288,1121,1112,925,739,724,688,654,655,644 |
| BRCA2 | single base substitution | 604,613,868,874,878,1143,1158,1159,1172,1180,1190,1191,1192,1257,1268,1338,1346,1350,1352,1354,1355,1512,1514,1529,1533,1569,1570,1593,1597,2336,2415,2418,2420,2422,2635,2650,2835,2837,2841,2842,2926,3049,3052,3085,3090,3095,3103,3177,3194,3297,3304,3308,3319,3322,3 |
| BRCA2 | insertion/deletion | 260,599,605,885,1200,1258,1270,1353,1362,1364,1517,3069,3172,3278, |
| BRIP1 | single base substitution | 988,984,919,918,913,910,722,718,717,470,453,439,421,416,404,403, |
| BRIP1 | insertion/deletion | 540, |
| C11orf30 | single base substitution | 263,267,277,376,394,410,415,434,439,445,539,943,950,952,1148,1150,11 |
| C11orf30 | insertion/deletion | 552, |
| CDH1 | single base substitution | 353, 370, 402 |
| CDH1 | insertion/deletion | 82, 343, 399 |
| CDKN2A | single base substitution | 51, 52, 57, 58, 60, 61, 63, 64, 65, 66, 67, 68, 69, 70, 71, 72, 74, 75, 76, 79, 80, 81, 82, 83, 84, 100, 101, 102, 106, 108, 110, 112, 114, 115, 116, 119, 120, 122, 123, 124, 125, 126, 128, 129, 130, 131, 132, 135 |
| CDKN2A | insertion/deletion | 53-58, 59, 76, 77, 78, 81, 84, 104, 105, 122, 124 |
| CHEK1 | single base substitution | 1,46,63,119,122,130,141,160,166,169,223,225,233,235,239,249,289,309,314,320,322,352,360,364,419,439,464,470, |
| CHEK1 | insertion/deletion | 99,226,234, |
| CHEK2 | single base substitution | 363,359,355,354,351,346,331,321,308,306,305,210,203, |
| CSF1R | single base substitution | 301, 969 |
| CTNNB1 | single base substitution | 13, 15, 20, 21, 22, 23, 25, 29, 30, 31, 32, 33, 34, 35, 36, 37, 38, 39, 40, 41, |
| CTNNB1 | insertion/deletion | 22-38, 42 |
| EGFR | single base substitution | 108, 289, 596, 598, 698, 702, 703, 709, 712, 719, 720, 721, 724, 730, 731, 733, 734, 735, 738, 741, 742, 743, 745, 746, 747, 750, 751, 752, 753, 754, 761, 765, 767, 768, 769, 770, 773, 774, 775, 776, 779, 784, 786, 790, 792, 810, 814, 819, 821, 858, 859, 861, 863, 864, 868, 870, 871, 873, 874 |
| EGFR | insertion/deletion | 745-759, 769-774 |
| ERBB2 | single base substitution | 755, 767, 769, 776, 777, 842, 878 |
| ERBB2 | insertion/deletion | 774-781 |

| | | |
|---|---|---|
| ERBB3 | single base substitution | 84,91,94,101,104,297,298,330,340,345,355,378,388,528,667,669,678,783, 792,1169,1173,1255,1261, |
| ERBB3 | insertion/deletion | 805, |
| ERBB4 | single base substitution | 140, 181, 244, 285, 303, 306, 341, 348, 595, 618, 931, 935, 936 |
| ESR1 | single base substitution | 6,9,20,29,40,218,222,227,235,440,443,448,457,533,537,538,541,545,548, |
| ESR1 | insertion/deletion | 29,451, |
| FANCA | single base substitution | 1094,1088,1084,1077,1075,1067,1053,1039,1038,1015,1011,1003,998,949,944,932,809,808,804,168,163,160,52,51,47,46,40, |
| FANCA | insertion/deletion | 1320,772, |
| FANCC | single base substitution | 22,20, |
| FANCD2 | single base substitution | 53,56,432,433,440,679,682,685,699,702,727,731,735,766,779,785,1042,1055,1123,1125,1126,1134,1149, |
| FANCE | single base substitution | 163,225,233,236,241,251,257,265, |
| FANCE | insertion/deletion | 246, |
| FANCF | single base substitution | 187,186,185,176,171, |
| FANCG | single base substitution | 471,468,451,450,163,161,156,155,135, |
| FANCL | single base substitution | 71,68, |
| FBXW7 | single base substitution | 226, 266, 278, 385, 393, 425, 465, 479, 504, 505, 582 |
| FGFR1 | single base substitution | 125, 252 |
| FGFR2 | single base substitution | 252, 253, 310, 372, 375, 382, 549 |
| FGFR3 | single base substitution | 248, 249, 251, 369, 370, 371, 373, 380, 384, 391, 641, 650, 697, 794 |
| FLT3 | single base substitution | 451, 572, 592, 599, 680, 831, 834, 835, 836, 837, 842 |
| FLT3 | insertion/deletion | 594-604, 835, 836, 840-841 |
| GATA3 | single base substitution | 263,269,276,279,294,299,300,309,313,316,317,325,326,331,336,344,365,367,369,371,377,391,393,396,403,404,408,409,416,417,419,422,436,443, |
| GATA3 | insertion/deletion | 309,315,316,318,321,327,331,330,332,333,334,336,345,346,351,352,358,357,360,365,363,379,388,395,399,403,401,402,408,409,411,419,420,426,427,428,433,434,435,437,439,440,442,443, |
| GNAS | single base substitution | 201, 227, 233, 844, 876 |
| HRAS | single base substitution | 11, 12, 13, 27, 61, 62 |
| IDH1 | single base substitution | 105, 116, 119, 123, 130, 132, 133, 134 |
| JAK2 | single base substitution | 611, 617, 618, 620 |
| KDR | single base substitution | 248, 275, 476, 873, 973, 984, 1140, 1145, 1210, 1308 |
| KDR | insertion/deletion | 1355 |

| Gene | Type | Positions |
|------|------|-----------|
| KIT | single base substitution | 51, 52, 509, 530, 541, 546, 551, 552, 553, 554, 555, 557, 558, 559, 560, 561, 562, 566, 567, 568, 572, 573, 576, 577, 582, 584, 585, 642, 643, 654, 670, 811, 816, 818, 820, 822, 823, 825, 839, 853 |
| KIT | insertion/deletion | 579, 715, 503-504, 551-562, 563-576 |
| KRAS | single base substitution | 8, 11, 12, 13, 14, 15, 18, 19, 22, 58, 59, 61, 117, 146 |
| KRAS | insertion/deletion | 13-Oct |
| MAP2K4 | single base substitution | 52,79,80,84,85,96,99,104,110,112,123,125,134,137,141,142,151,154,165, 184,186,197,200,203,207,215,219,221,226,228,233,234,238,248,249,251, 252,257,258,265,271,273,274,277,279,280,281,287,290,291,299,303,304, 306,309,310,316,320,321,322,326,327,341,348,349,364,373,378,385,389, |
| MAP2K4 | insertion/deletion | 113,149,160,173,190,223,263,266,295,319,346, |
| MAP3K1 | single base substitution | 5,31,38,39,45,117,126,143,149,155,188,192,195,201,229,233,260,268,362 ,369,384,406,438,443,509,527,536,537,540,547,551,596,607,609,613,627, 655,662,663,676,690,692,787,791,806,813,814,818,821,857,861,885,913, 914,962,965,969,1003,1019,1021,1058,1061,1066,1074,1167,1180,1211,1 |
| MAP3K1 | insertion/deletion | 9,48,59,70,110,123,146,156,162,191,193,208,234,294,407,425,509,514,55 2,600,601,616,633,675,695,766,787,803,837,875,877,878,914,962,982,98 5,1053,1070,1084,1127,1183,1222,1241,1245,1250,1265,1311,1323,1329, |
| MAP3K4 | single base substitution | 362,439,447,483,486,491,835,1183,1199,1209,1608, |
| MAP3K4 | insertion/deletion | 453,471,1199, |
| MET | single base substitution | 168, 375, 848, 988, 1010, 1112, 1117, 1118, 1124, 1130, 1248, 1253, 1268 |
| MET | insertion/deletion | 982-1028 |
| MLH1 | single base substitution | 384 |
| MPL | single base substitution | 505, 506, 515, 519 |
| MYC | single base substitution | 404,405,416, |
| MYC | insertion/deletion | 392, |
| NBN | single base substitution | 748,740,729,714,712,710,688,685,684,679,664,661,660,646,637,636,624, 561,543,526,512,499,491,486,485,447,432,418,377,372,363,356,354,351, 346,338,327,322,302,284,272,268,242,235,224,222,218,195,194,186,168, |
| NBN | insertion/deletion | 731,654,551,466,125,103, |
| NOTCH1 | single base substitution | 1575, 1577, 1586, 1593, 1594, 1597, 1599, 1601, 1677, 1679, 1579, 2441, |
| NOTCH1 | insertion/deletion | 1579, 2463, 2464, 2468 |
| NPM1 | single base substitution | 290 |
| NPM1 | insertion/deletion | 288, 289, 290 |

| | | |
|---|---|---|
| NRAS | single base substitution | 11, 12, 13, 18, 60, 61, 65, 146 |
| PDGFRA | single base substitution | 561, 659, 674, 824, 842, 846 |
| PDGFRA | insertion/deletion | 554-555, 560-564, 566, 841-847 |
| PIK3CA | single base substitution | 60, 75, 80, 81, 88, 108, 110, 111, 112, 115, 324, 344, 345, 405, 407, 418, 420, 453, 457, 466, 539, 542, 544, 545, 546, 594, 701, 901, 909, 1020, 1021, 1023, 1025, 1029, 1035, 1039, 1040, 1043, 1044, 1045, 1046, 1047, |
| PIK3CA | insertion/deletion | 108, 109, 111-115, 451-456, 1068, 1069 |
| PIK3R1 | single base substitution | 13,22,406,408,409,419,449,450,452,454,455,457,458,459,461,462,463,464,466,470,495,496,498,503,510,514,515,519,522,557,559,560,561,564,565,567,568,570,571,572,573,574,576,577,580,627,634,635,642,649, |
| PIK3R1 | insertion/deletion | 398,401,405,404,406,419,446,449,450,451,452,453,454,455,456,457,458,462,459,460,465,463,464,468,466,434,511,556,558,559,560,562,564,563,567,568,570,571,573,574,575,576,577,578,579,580,581,582,633,636,642, |
| PTEN | single base substitution | 7, 8, 10, 13, 14, 15, 16, 17, 56, 57, 59, 61, 65,66, 68, 105, 107, 108, 110, 111, 112, 121, 123, 124, 125, 126, 127, 128, 129, 130, 132, 133, 165, 167, 170, 171, 173, 174, 177, 214, 235, 242, 245, 246, 248, 251, 252, 253, 256, |
| PTEN | insertion/deletion | 6, 16, 17, 57, 60, 63, 68, 108-109, 116, 118, 119, 122, 124, 128-130, 131, 173,176, 237, 242, 246, 248, 250, 254, 256, 260, 267, 288, 290, 293, 294, |
| PTPN11 | single base substitution | 52, 58, 60, 61, 69, 71, 72, 73, 76, 491, 502, 503, 510 |
| RAD50 | single base substitution | 292,305,315,318,355,358,362,366,384, |
| RAD50 | insertion/deletion | 300, |
| RAD51 | single base substitution | 86,218, |
| RB1 | single base substitution | 137, 199, 320, 358, 455, 552, 556, 579, 661, 685, 706, 715, 748, 754 |
| RB1 | insertion/deletion | 660, 676 |
| RET | single base substitution | 618, 620, 630, 631, 634, 768, 883, 918 |
| RET | insertion/deletion | 612-620, 629-640 |
| SLC7A5 | single base substitution | 247,244,231,229,227, |
| SMAD4 | single base substitution | 118, 144, 168, 245, 311, 330, 343, 351, 353, 355, 358, 361, 386, 412, 445, 497, 499, 507, 515, 523, 526 |
| SMAD4 | insertion/deletion | 123, 259, 339-343, 410,  448, 466, 517 |
| SMARCB1 | single base substitution | 40, 47, 53, 158, 201, 377 |
| SMARCB1 | insertion/deletion | 382, 383 |
| SMO | single base substitution | 199, 324, 404, 535, 640 |
| SRC | single base substitution | 531 |

| STK11 | single base substitution | 32, 37, 60, 159, 170, 171, 194, 196, 199, 272, 281, 332, 354 |
|---|---|---|
| STK11 | insertion/deletion | 57, 60, 264, 281, 282 |
| TP53 | single base substitution | 3, 11, 20, 25, 26, 31, 33, 41, 42, 43, 44, 47, 59, 60, 61, 64, 65, 66, 70, 72, 73, 75, 77, 78, 80, 81, 82, 83, 84, 85, 86, 87, 89, 91, 94, 95, 96, 98, 99, 100, 101, 102, 103, 104, 105, 106, 107, 108, 109, 110, 120, 126, 127, 128, 129, 130, 131, 132, 133, 134, 135, 136, 141, 143, 144, 145, 151, 152, 153, 154, 155, 156, 157, 158, 159, 160, 161, 162, 163, 164, 165, 166, 167, 168, 169, 170, 171, 172, 173, 174, 175, 176, 177, 178, 179, 180, 181, 182, 183, 184, 185, 186, 187, 189, 190, 191, 192, 193, 194, 195, 196, 197, 198, 199, 200, 201, 202, 203, 204, 205, 206, 207, 208, 209, 210, 211, 212, 213, 214, 215, 216, 217, 218, 219, 220, 221, 226, 227, 228, 229, 230, 231, 232, 233, 234, 235, 236, 237, 238, 239, 240, 241, 242, 243, 244, 245, 246, 247, 248, 249, 250, 251, 252, 253, 254, 255, 256, 265, 266, 267, 268, 269, 270, 271, 272, 273, 274, 275, 276, 277, 278, 279, 280, 281, 282, 283, 284, 285, 286, 287, |
| TP53 | insertion/deletion | 46, 59, 85, 76-90, 98, 99, 102, 105, 108, 126, 128, 131, 132, 133, 135, 154, 155, 156, 157, 158-159, 161, 162, 163, 164, 167, 171, 172, 173, 174, 175-182, 178, 185, 186, 189-197, 200, 201, 203, 204, 206, 209, 210, 211, 212, 213, 214, 215, 216, 218, 220, 229, 230, 231, 234, 235, 236, 237, 239, 240, 241, 242, 244, 245, 246, 247-248, 249, 250-253, 269-270, 272, 273, 278, |
| VHL | single base substitution | 81, 82, 85, 86, 88, 89, 90, 93, 95, 96, 98, 101, 114, 115, 117, 118, 119, 121, 122, 126, 128, 130, 131, 132, 134, 136, 138, 139, 143, 144, 146, 157, 158, 160, 161, 162, 163, 164, 166, 167, 169, 171 |
| VHL | insertion/deletion | 87-88, 90, 98, 99, 104, 115, 124, 128, 131, 133, 135, 136, 137, 138, 139, 141, 142, 144, 146, 148, 155, 156, 157, 160, 163, 167, 168 |