

Auflistung der 174 Genbereiche, die mit dem Actionable Solid Tumor Panel (Qiagen) untersucht werden; insgesamt 13.296 Basen kodierende Sequenz

| Gen | Lage (hg19) | Transkript | Amplifikat (Covered target region) | | | | | | Wichtige Mutationen (nach COSMIC) |
|-------------|-------------|-------------|------------------------------------|-------------|-------------|-------------|---------------------|--------------------------------|---|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | Größe nicht-kodierende Sequenz | |
| AKT1 | chr14 | NM_005163.2 | AKT1 Exon 3 | 105.246.541 | 105.246.562 | 16 - 20 | 22 | 9 | COSM33765: c.49G>A (E17K) |
| | | | | | | | 22 | 13 | Basen kodierende Sequenz gesamt |
| ALK | chr2 | NM_004304.4 | ALK Exon 5 | 29.606.669 | 29.606.689 | 398 - 404 | 21 | | COSM20713: c.1201C>T (R401*) |
| | | | ALK Exon 8 | 29.541.181 | 29.541.201 | 539 - 545 | 21 | | COSM48230: c.1625C>G (P542R) |
| | | | ALK Exon 15 | 29.455.257 | 29.455.277 | 842 - 848 | 21 | | COSM148825: c.2535T>C (G845G) |
| | | | ALK Exon 18 | 29.449.809 | 29.449.829 | 1009 - 1015 | 21 | | COSM148824: c.3036G>A (T1012T) |
| | | | ALK Exon 19 | 29.448.400 | 29.448.420 | 1027 - 1033 | 21 | | COSM4133807: c.3089A>C (H1030P) |
| | | | ALK Exon 20 | 29.446.286 | 29.446.306 | 1088 - 1094 | 21 | | COSM28502: c.3271G>A (D1091N) |
| | | | ALK Exon 22 | 29.445.246 | 29.445.283 | 1151 - 1160 | 38 | 9 | COSM98478: c.3452C>T (T1151M) |
| | | | ALK Exon 22 | 29.445.182 | 29.445.223 | 1168 - 1172 | 42 | 28 | COSM28498: c.3512T>A (I1171N) |
| | | | ALK Exon 23 | 29.443.684 | 29.443.707 | 1172 - 1178 | 24 | 6 | COSM28055: c.3522C>A (F1174L) |
| | | | ALK Exon 23 | 29.443.597 | 29.443.641 | 1193 - 1207 | 45 | | COSM99137: c.3586C>A (L1196M) |
| | | | ALK Exon 24 | 29.436.848 | 29.436.870 | 1242 - 1248 | 23 | 2 | COSM28499: c.3733T>G(F1245V) |
| | | | ALK Exon 25 | 29.432.645 | 29.432.693 | 1266 - 1279 | 49 | 7 | COSM28056: c.3824G>A (R1275Q) |
| | | | ALK Exon 29 | 29.416.605 | 29.416.625 | 1443 - 1449 | 21 | | COSM5019786: c.4338C>T (T1446T) |
| | | | ALK Exon 29 | 29.416.471 | 29.416.491 | 1488 - 1494 | 21 | | COSM1130802: c.4472A>G (K1491R) |

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|---------------|----------------|----------------|------------------------------------|-------------|-------------|-------------|------------------------|---------------------------------------|---|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | Größe nicht- kodierende Sequenz | |
| | | | | | | | 389 | 337 | Basen kodierende Sequenz gesamt |
| BRAF | chr7 | NM_004333.4 | BRAF Exon 7 | 140.500.155 | 140.500.175 | 323 - 327 | 21 | | COSM33964: c.977T>C (I326T) |
| | | | BRAF Exon 11* | 140.481.376 | 140.481.503 | 439 - 477 | 128 | 10 | COSM460: c.1406G>C (G469A) |
| | | | BRAF Exon 12* | 140.477.781 | 140.477.885 | 478 - 506 | 105 | 20 | COSM243226: c.1514T>A (L505H) |
| | | | BRAF Exon 15* | 140.453.065 | 140.453.203 | 581 - 620 | 139 | 20 | COSM476: c.1799T>A (V600E) |
| | | | BRAF Exon 16 | 140.449.140 | 140.449.180 | 634 - 646 | 41 | | COSM4161857: c.1929A>G (G643G) |
| | | | | | | | 434 | 384 | Basen kodierende Sequenz gesamt |
| CTNNB1 | chr3 | NM_001098209.1 | CTNNB1 Exon 3* | 41.266.007 | 41.266.254 | 5 - 81 | 248 | 20 | COSM5664: c.121A>G (T41A); COSM5667: c.134C>T (S45F) |
| | | | | | | | 248 | 228 | Basen kodierende Sequenz gesamt |
| EGFR | chr7 | NM_005228.3 | EGFR Exon 2 | 55.210.065 | 55.210.087 | 59 - 66 | 23 | | COSM35602: c.185T>G (L62R) |
| | | | EGFR Exon 3 | 55.211.070 | 55.211.107 | 105 - 117 | 38 | | COSM21683: c.323G>A (R108K) |
| | | | EGFR Exon 4 | 55.214.338 | 55.214.358 | 155 - 161 | 21 | | COSM42978: c.474C>T (N158N) |
| | | | EGFR Exon 6 | 55.220.264 | 55.220.284 | 219 - 225 | 21 | | COSM35508: c.664C>T (R222C) |
| | | | EGFR Exon 7 | 55.221.700 | 55.221.720 | 250 - 255 | 21 | 4 | COSM35752: c.754C>T (R252C) |
| | | | EGFR Exon 7 | 55.221.733 | 55.221.753 | 260 - 266 | 21 | | COSM21684: c.787A>C (T263P) |
| | | | EGFR Exon 7 | 55.221.811 | 55.221.832 | 286 - 292 | 22 | | COSM21687: c.866C>T (A289V) |

| Gen | Lage (hg19) | Transkript | Amplifikat (Covered target region) | | | | | Wichtige Mutationen (nach COSMIC) | |
|--------------|----------------|---------------------|------------------------------------|------------|------------|------------------------------|------------------------|--------------------------------------|--|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | | Größe nicht- kodierende Sequenz |
| | | | EGFR Exon 8 | 55.223.533 | 55.223.553 | 301 - 307 | 21 | | COSM2149971: c.910C>T (H304Y) |
| | | | EGFR Exon 12 | 55.227.997 | 55.228.019 | 489 - 495 | 23 | | COSM236670: c.1476C>A (S492R) |
| | | | EGFR Exon 13 | 55.229.245 | 55.229.265 | 518 - 524 | 21 | | COSM3721608: c.1562G>A (R521K) |
| | | | EGFR Exon 15 | 55.233.027 | 55.233.053 | 593 - 601 | 27 | | COSM21690: c.1793G>T (G598V) |
| | | | EGFR Exon 15 | 55.233.099 | 55.233.119 | 617 - 623 | 21 | | COSM35825: c.1859G>A (C620Y) |
| | NM_201284 | <i>EGFR Exon 16</i> | 55.238.077 | 55.238.097 | 653 - 659 | 21 | | COSM54529: c.1957A>T (S653C) | |
| | NM_201284 | <i>EGFR Exon 16</i> | 55.238.102 | 55.238.122 | 662 - 668 | 21 | | COSM3942265: c.1993G>T (E665*) | |
| | | | EGFR Exon 17 | 55.240.697 | 55.240.717 | 648 - 654 | 21 | | COSM35840: c.1951G>A (V651M) |
| | | | EGFR Exon 17 | 55.240.776 | 55.240.816 | 674 - 687 | 41 | | COSM22555: c.2030G>A (R677H) |
| | | | EGFR Exon 18* | 55.241.617 | 55.241.637 | 689 - 695 | 21 | | COSM13178: c.2081C>T (P694L) |
| | | | EGFR Exon 18* | 55.241.646 | 55.241.736 | 699 - 728 | 91 | | COSM6239: c.2156G>C (G719A) |
| | | | <i>Intron 18-19</i> | 55.241.745 | 55.241.765 | | 21 | 21 | |
| | | | EGFR Exon 19* | 55.242.405 | 55.242.523 | 729 - 761 | 119 | 20 | COSM6223: c.2235_2249del15 (E746_A750del) |
| | | | EGFR Exon 20* | 55.248.976 | 55.249.181 | 762 - 823 | 206 | 20 | COSM6240: c.2369C>T (T790M) |
| | | | EGFR Exon 21* | 55.259.402 | 55.259.577 | 824 - 875 | 176 | 20 | COSM6224: c.2573T>G (L858R) |
| | | | EGFR Exon 22 | 55.260.473 | 55.260.493 | 881 - 887 | 21 | | COSM28575: c.2650G>A (E884K) |
| EGFR Exon 23 | 55.266.459 | 55.266.481 | 918 - 924 | 23 | | COSM28515: c.2763C>A (S921R) | | | |

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|--------------|----------------|-------------|------------------------------------|------------|------------|-------------|------------------------|---------------------------------------|---------------------------------------|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | Größe nicht- kodierende Sequenz | |
| | | | EGFR Exon 25 | 55.268.906 | 55.268.926 | 991 - 997 | 21 | | COSM3762773: c.2982C>T (D994D) |
| | | | | | | | 1083 | 998 | Basen kodierende Sequenz gesamt |
| ERBB2 | chr17 | NM_004448.2 | <i>Intron</i> | 37.855.803 | 37.855.850 | | 48 | 48 | |
| | | | ERBB2 Exon 1* | 37.856.482 | 37.856.574 | 1 - 25 | 93 | 20 | COSM5608588: c.55G>A (G19R) |
| | | | ERBB2 Exon 2* | 37.863.233 | 37.863.404 | 25 - 75 | 172 | 20 | COSM5962860: c.106C>T (P36S) |
| | | | ERBB2 Exon 3* | 37.864.564 | 37.864.797 | 76 - 147 | 234 | 20 | COSM6939834: c.308G>A (R103Q) |
| | | | ERBB2 Exon 4* | 37.865.561 | 37.865.715 | 147 - 192 | 155 | 20 | COSM5458623: c.497C>T (T166M) |
| | | | ERBB2 Exon 5* | 37.866.056 | 37.866.144 | 192 - 215 | 89 | 20 | COSM5715624: c.619G>A (E207K) |
| | | | ERBB2 Exon 6* | 37.866.329 | 37.866.464 | 215 - 253 | 136 | 20 | COSM3387843: c.649C>T (R217C) |
| | | | ERBB2 Exon 7* | 37.866.583 | 37.866.744 | 254 - 301 | 162 | 20 | COSM4767524: c.874G>C (G292R) |
| | | | ERBB2 Exon 8* | 37.868.171 | 37.868.310 | 301 - 341 | 140 | 20 | COSM48358: c.929C>T (S310F) |
| | | | ERBB2 Exon 9* | 37.868.565 | 37.868.711 | 341 - 383 | 147 | 20 | COSM6906934: c.1110C>G (I370M) |
| | | | ERBB2 Exon 10* | 37.871.529 | 37.871.622 | 383 - 408 | 94 | 20 | COSM4600589: c.1177C>A (Q393K) |
| | | | ERBB2 Exon 11* | 37.871.689 | 37.871.799 | 408 - 438 | 111 | 20 | COSM3387846: c.1270G>A (V424I) |
| | | | ERBB2 Exon 12* | 37.871.983 | 37.872.202 | 438 - 505 | 220 | 20 | COSM6005786: c.1436C>T (T479M) |
| | | | ERBB2 Exon 13* | 37.872.544 | 37.872.696 | 505 - 549 | 153 | 20 | COSM5962315: c.1634G>A (R545Q) |
| | | | ERBB2 Exon 14* | 37.872.758 | 37.872.868 | 549 - 579 | 111 | 20 | COSM6865878: c.1702C>T (Q568*) |

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|--------------|----------------|-------------|------------------------------------|------------|------------|--------------|------------------------|--------------------------------------|--|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | | Größe nicht- kodierende Sequenz |
| | | | ERBB2 Exon 15* | 37.873.563 | 37.873.743 | 580 - 633 | 181 | 20 | COSM6904430: c.1880C>A (P627H) |
| | | | ERBB2 Exon 16* | 37.876.030 | 37.876.097 | 633 - 649 | 68 | 20 | COSM6008914: c.1898C>T (S633F) |
| | | | ERBB2 Exon 17* | 37.879.562 | 37.879.720 | 649 - 695 | 159 | 20 | COSM436498: c.2033G>A (R678Q); COSM4000121: c.1963A>G (I655V) |
| | | | ERBB2 Exon 18* | 37.879.781 | 37.879.923 | 696 - 736 | 143 | 20 | COSM14059: c.2198C>T (T733I) |
| | | | ERBB2 Exon 19* | 37.880.155 | 37.880.273 | 737 - 769 | 119 | 20 | COSM14060: c.2264T>C (L755S); COSM1251412: c.2305G>T (D769Y) |
| | | | ERBB2 Exon 20* | 37.880.969 | 37.881.174 | 770 - 831 | 206 | 20 | COSM14062: c.2329G>T (V777L); COSM20959: c.2324_2325ins12 |
| | | | ERBB2 Exon 21* | 37.881.292 | 37.881.467 | 832 - 883 | 176 | 20 | COSM14065: c.2524G>A (V842I) |
| | | | ERBB2 Exon 22* | 37.881.570 | 37.881.665 | 884 - 909 | 96 | 20 | COSM978678: c.2690G>A (R897Q) |
| | | | ERBB2 Exon 23* | 37.881.950 | 37.882.116 | 909 - 958 | 167 | 20 | COSM6005645: c.2790G>T (E930D) |
| | | | ERBB2 Exon 24* | 37.882.805 | 37.882.922 | 958 - 990 | 118 | 20 | COSM88181: c.2912A>G (E971G) |
| | | | ERBB2 Exon 25* | 37.883.058 | 37.883.266 | 991 - 1053 | 209 | 20 | COSM5962765: c.3097C>G (P1033A) |
| | | | ERBB2 Exon 26* | 37.883.538 | 37.883.810 | 1054 - 1138 | 273 | 20 | COSM3378169: c.3382G>A (V1128I) |
| | | | ERBB2 Exon 27* | 37.883.932 | 37.884.307 | 1138 - 1256* | 376 | 20 | COSM5436892: c.3508C>G (P1170A) |
| | | | | | | | 4356 | 3768 | Basen kodierende Sequenz gesamt |
| ERBB3 | chr12 | NM_001982.3 | ERBB3 Exon 2 | 56.477.621 | 56.477.641 | 57 - 63 | 21 | | COSM1251422: c.179T>A (M60K) |
| | | | ERBB3 Exon 3 | 56.478.844 | 56.478.864 | 101 - 107 | 21 | | COSM20710: c.310G>A (V104M) |

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|--------------|----------------|-------------|------------------------------------|-------------|-------------|-------------|------------------------|---------------------------------------|---|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | Größe nicht- kodierende Sequenz | |
| | | | ERBB3 Exon 7 | 56.481.912 | 56.481.932 | 281 - 287 | 21 | | COSM48360: c.850G>A (G284R) |
| | | | ERBB3 Exon 8 | 56.482.331 | 56.482.351 | 294 - 298 | 21 | | COSM1299647: c.889G>T (D297Y) |
| | | | ERBB3 Exon 9 | 56.482.527 | 56.482.547 | 330 - 335 | 21 | 5 | COSM1747136: c.994G>A (E332K) |
| | | | ERBB3 Exon 12 | 56.487.191 | 56.487.211 | 446 - 452 | 21 | | COSM5019625: c.1347T>C (I449I) |
| | | | ERBB3 Exon 23 | 56.492.623 | 56.492.643 | 925 - 931 | 21 | | COSM1363010: c.2783A>G (E928G) |
| | | | ERBB3 Exon 27 | 56.494.988 | 56.495.008 | 1116 - 1122 | 21 | | COSM4989443: c.3355A>T (S1119C) |
| | | | | | | | 168 | 163 | Basen kodierende Sequenz gesamt |
| ESR1 | chr6 | NM_000125.3 | ESR1 Exon 1 | 152.129.067 | 152.129.087 | 7 - 13 | 21 | | COSM3761554: c.30T>C (S10S) |
| | | | ESR1 Exon 4 | 152.265.445 | 152.265.465 | 300 - 306 | 21 | | COSM4745827: c.908A>G (K303R) |
| | | | <i>Intron</i> | 152.396.026 | 152.396.046 | | 21 | 21 | |
| | | | ESR1 Exon 8 | 152.419.912 | 152.419.936 | 534 - 541 | 25 | | COSM94250: c.1613A>G (D538G); COSM1074639: c.1610A>C (Y537S) |
| | | | ESR1 Exon 8 | 152.420.085 | 152.420.105 | 591 - 596* | 21 | 4 | COSM3761556: c.1782G>A (T594T) |
| | | | | | | | 109 | 84 | Basen kodierende Sequenz gesamt |
| FOXL2 | chr3 | NM_023067.3 | FOXL2 Exon 1 | 138.665.153 | 138.665.174 | 131 - 137 | 22 | | COSM33661: c.402C>G (C134W) |
| | | | | | | | 22 | 22 | Basen kodierende Sequenz gesamt |
| GNA11 | chr19 | NM_002067.2 | GNA11 Exon 5 | 3.118.932 | 3.118.953 | 206 - 212 | 22 | | COSM52969: c.626A>T (Q209L) |

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|-------------|----------------|-------------|------------------------------------|-------------|-------------|-------------|------------------------|---------------------------------------|---|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | Größe nicht- kodierende Sequenz | |
| | | | | | | | 22 | 22 | Basen kodierende Sequenz gesamt |
| GNAQ | chr9 | NM_002072.4 | GNAQ Exon 5 | 80.409.478 | 80.409.500 | 205 - 212 | 23 | | COSM28757: c.626A>T (Q209L); COSM28758: c.626A>C (Q209P) |
| | | | | | | | 23 | 23 | Basen kodierende Sequenz gesamt |
| IDH1 | chr2 | NM_005896.3 | IDH1 Exon 4 | 209.113.103 | 209.113.124 | 128 - 135 | 22 | | COSM28746: c.395G>A (R132H); COSM28747: c.394C>T (R132C) |
| | | | | | | | 22 | 22 | Basen kodierende Sequenz gesamt |
| IDH2 | chr15 | NM_002168.2 | IDH2 Exon 4 | 90.631.828 | 90.631.849 | 169 - 175 | 22 | | COSM33733: c.515G>A (R172K) |
| | | | IDH2 Exon 4 | 90.631.924 | 90.631.945 | 137 - 143 | 22 | | COSM41590: c.419G>A (R140Q) |
| | | | | | | | 44 | 44 | Basen kodierende Sequenz gesamt |
| KIT | chr4 | NM_000222.2 | KIT Exon 2 | 55.561.748 | 55.561.775 | 47 - 55 | 28 | | COSM1146: c.154G>A (D52N) |
| | | | KIT Exon 8 | 55.589.758 | 55.589.788 | 414 - 423 | 31 | | COSM29014: c.1255_1257delGAC (D419del) |
| | | | KIT Exon 9* | 55.592.013 | 55.592.226 | 449 - 514 | 214 | 20 | COSM1326: c.1509_1510insGCCTAT (Y503_F504insAY) |
| | | | KIT Exon 10 | 55.593.454 | 55.593.491 | 538 - 549 | 38 | 1 | COSM28026: c.1621A>C (M541L) |
| | | | KIT Exon 11* | 55.593.572 | 55.593.718 | 550 - 592 | 147 | 20 | COSM1217: c.1669_1674del6 (W557_K558del) |
| | | | KIT Exon 13* | 55.594.167 | 55.594.297 | 627 - 664 | 131 | 20 | COSM1304: c.1924A>G (K642E), COSM12706: c.1961T>C (V654A) |
| | | | KIT Exon 14* | 55.595.491 | 55.595.661 | 664 - 714 | 171 | 20 | COSM12708: c.2009C>T (T670I) |
| | | | KIT Exon 15 | 55.597.485 | 55.597.507 | 714 - 718 | 23 | 9 | COSM1306: c.2143_2145delAGC (S715del) |

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|-------------|----------------|----------------|------------------------------------|-------------|-------------|-------------|------------------------|--------------------------------------|---------------------------------------|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | | |
| | | | <i>Intron</i> | 55.599.149 | 55.599.169 | | 21 | 21 | |
| | | | KIT Exon 17* | 55.599.226 | 55.599.368 | 788 - 828 | 143 | 20 | COSM1314: c.2447A>T (D816V) |
| | | | <i>Intron</i> | 55.599.391 | 55.599.411 | | 21 | 21 | |
| | | | KIT Exon 18* | 55.602.654 | 55.602.674 | 829 - 832 | 21 | 10 | COSM13172: c.2485G>C (A829P) |
| | | | KIT Exon 18* | 55.602.684 | 55.602.704 | 836 - 842 | 21 | | COSM1324: c.2515G>A (E839K) |
| | | | KIT Exon 18* | 55.602.753 | 55.602.775 | 859 - 865 | 23 | | COSM1325: c.2586G>C (L862L) |
| | | | KIT Exon 20 | 55.603.362 | 55.603.382 | 907 - 913 | 21 | | COSM5609257: c.2732C>T (P911L) |
| | | | KIT Exon 21 | 55.604.649 | 55.604.669 | 953 - 959 | 21 | | COSM71350: c.2867G>A (R956Q) |
| | | | | | | | 1075 | 913 | Basen kodierende Sequenz gesamt |
| KRAS | chr12 | NM_004985.3 | KRAS 3'UTR | 25.360.214 | 25.360.234 | | 21 | 21 | |
| | | | KRAS Exon 5 | 25.362.795 | 25.362.815 | 161 - 167 | 21 | | COSM41307: c.491G>A (p.R164Q) |
| | | | KRAS Exon 4* | 25.378.538 | 25.378.717 | 97 - 150 | 180 | 20 | COSM19404: c.436G>A (A146T) |
| | | | KRAS Exon 3* | 25.380.158 | 25.380.356 | 38 - 97 | 199 | 20 | COSM554: c.183A>C (Q61H) |
| | | | KRAS Exon 2* | 25.398.198 | 25.398.339 | 1 - 37 | 142 | 20 | COSM521: c.35G>A (G12D) |
| | | | | | | | 563 | 482 | Basen kodierende Sequenz gesamt |
| MET | chr7 | NM_001127500.2 | MET Exon 19 | 116.423.397 | 116.423.418 | 1243 - 1249 | 22 | | COSM699: c.3743A>G (Y1248C) |

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|---------------|----------------|-------------|------------------------------------|-------------|-------------|-------------|------------------------|---------------------------------------|---|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | Größe nicht- kodierende Sequenz | |
| | | | MET Exon 19 | 116.423.464 | 116.423.485 | 1265 - 1271 | 22 | | COSM691: c.3803T>C (M1268T) |
| | | | | | | | 44 | 44 | Basen kodierende Sequenz gesamt |
| NRAS | chr1 | NM_002524.4 | NRAS Exon 4* | 115.252.180 | 115.252.359 | 97 - 150 | 180 | 20 | COSM4170228: c.437C>T (A146V) |
| | | | NRAS Exon 3* | 115.256.411 | 115.256.609 | 38 - 97 | 199 | 20 | COSM584: c.182A>G (Q61R) |
| | | | NRAS Exon 2* | 115.258.661 | 115.258.808 | 1 - 37 | 148 | 20 | COSM564: c.35G>A (G12D) |
| | | | | | | | 527 | 467 | Basen kodierende Sequenz gesamt |
| PDGFRA | chr4 | NM_006206.4 | PDGFRA Exon 3 | 55.127.438 | 55.127.458 | 76 - 82 | 21 | | COSM5019287: c.236G>A (G79D) |
| | | | PDGFRA Exon 4 | 55.130.068 | 55.130.088 | 201 - 207 | 21 | | COSM4416371: c.612T>C (N204N) |
| | | | PDGFRA Exon 5 | 55.131.108 | 55.131.128 | 218 - 224 | 21 | | COSM6438148: c.661C>T (L221F) |
| | | | PDGFRA Exon 5 | 55.131.132 | 55.131.171 | 226 - 238 | 40 | | COSM42897: c.704G>A (C235Y) |
| | | | PDGFRA Exon 7 | 55.133.716 | 55.133.736 | 311 - 316 | 21 | 3 | COSM1666925: c.939T>G (G313G) |
| | | | PDGFRA Exon 10 | 55.139.761 | 55.139.781 | 475 - 481 | 21 | | COSM5008347: c.1432T>C (S478P) |
| | | | PDGFRA Exon 12* | 55.140.998 | 55.141.150 | 552 - 596 | 153 | 20 | COSM12418: c.1698_1712del15; COSM739: c.1682T>A (V561D) |
| | | | PDGFRA Exon 13 | 55.143.567 | 55.143.587 | 600 - 606 | 21 | | COSM4417622: c.1809G>A (A603A) |
| | | | PDGFRA Exon 14* | 55.144.053 | 55.144.183 | 631 - 668 | 131 | 20 | COSM22414: c.1977C>G (N659K) |
| | | | PDGFRA Exon 15 | 55.144.537 | 55.144.557 | 671 - 677 | 21 | | COSM743: c.2021C>T (T674I) |

| Gen | Lage (hg19) | Transkript | Amplifikat (Covered target region) | | | | | Wichtige Mutationen (nach COSMIC) | |
|-----------------|----------------|-------------|------------------------------------|-------------|-------------|-------------------------------|------------------------|--------------------------------------|---|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | | Größe nicht- kodierende Sequenz |
| | | | PDGFRA Exon 18* | 55.151.998 | 55.152.140 | 814 - 854 | 143 | 20 | COSM736: c.2525A>T (D842V) ; COSM22413: c.2472C>T (V824V) |
| | | | <i>Intron</i> | 55.153.537 | 55.153.558 | | 22 | 22 | |
| | | | PDGFRA Exon 23 | 55.161.370 | 55.161.390 | 1068 - 1074 | 21 | | |
| | | | | | | | 657 | 572 | Basen kodierende Sequenz gesamt |
| PIK3CA | chr3 | NM_006218.2 | PIK3CA Exon 2* | 178.916.604 | 178.916.975 | 1 - 118 | 372 | 20 | COSM746: c.263G>A (R88Q) |
| | | | PIK3CA Exon 3* | 178.917.468 | 178.917.697 | 118 - 188 | 230 | 20 | COSM751: c.353G>A (G118D) |
| | | | PIK3CA Exon 4* | 178.919.068 | 178.919.338 | 188 - 271 | 271 | 20 | COSM86047: c.665C>T (A222V) |
| | | | PIK3CA Exon 5* | 178.921.322 | 178.921.587 | 272 - 353 | 266 | 20 | COSM754: c.1035T>A (N345K) |
| | | | PIK3CA Exon 6* | 178.922.281 | 178.922.386 | 354 - 382 | 106 | 20 | COSM86044: c.1093G>A (E365K) |
| | | | PIK3CA Exon 7* | 178.927.373 | 178.927.498 | 382 - 417 | 126 | 20 | COSM328028: c.1173A>G (I391M) |
| | | | PIK3CA Exon 8* | 178.927.964 | 178.928.136 | 418 - 468 | 173 | 20 | COSM757: c.1258T>C (C420R) |
| | | | PIK3CA Exon 9* | 178.928.209 | 178.928.363 | 469 - 513 | 155 | 20 | COSM136387: c.1412C>T (P471L) |
| | | | PIK3CA Exon 10* | 178.935.988 | 178.936.132 | 514 - 555 | 145 | 20 | COSM763: c.1633G>A (E545K) ; COSM760: c.1624G>A (E542K) |
| | | | PIK3CA Exon 11* | 178.936.974 | 178.937.075 | 555 - 582 | 102 | 20 | COSM30790: c.1700A>G (K567R) |
| | | | PIK3CA Exon 12* | 178.937.349 | 178.937.533 | 583 - 637 | 185 | 20 | COSM1041500: c.1810T>C (C604R) |
| PIK3CA Exon 13* | 178.937.727 | 178.937.850 | 638 - 672 | 124 | 20 | COSM250052: c.1913T>C (V638A) | | | |

| Gen | Lage (hg19) | Transkript | Amplifikat (Covered target region) | | | | | | Wichtige Mutationen (nach COSMIC) |
|-------------|----------------|-------------|------------------------------------|-------------|-------------|-------------|------------------------|---------------------------------------|--|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | Größe nicht- kodierende Sequenz | |
| | | | PIK3CA Exon 14* | 178.938.764 | 178.938.955 | 672 - 729 | 192 | 20 | COSM87306: c.2176G>A (E726K) |
| | | | PIK3CA Exon 15* | 178.941.859 | 178.941.985 | 730 - 765 | 127 | 20 | COSM6148: c.2198A>G (K733R) |
| | | | PIK3CA Exon 16* | 178.942.478 | 178.942.619 | 765 - 806 | 142 | 20 | COSM4598858: c.2352G>C (E784D) |
| | | | PIK3CA Exon 17* | 178.943.740 | 178.943.838 | 806 - 832 | 99 | 20 | COSM1041508: c.2452C>T (R818C) |
| | | | PIK3CA Exon 18* | 178.947.050 | 178.947.240 | 832 - 889 | 191 | 20 | COSM4481256: c.2494C>T (R832*) |
| | | | PIK3CA Exon 19* | 178.947.782 | 178.947.919 | 889 - 928 | 138 | 20 | COSM769: c.2702G>T (C901F) |
| | | | PIK3CA Exon 20* | 178.948.003 | 178.948.174 | 929 - 979 | 172 | 20 | COSM94980: c. 2908G>A (E970K) |
| | | | PIK3CA Exon 21* | 178.951.872 | 178.952.162 | 979 - 1069* | 291 | 20 | COSM775: c.3140A>G (H1047R) |
| | | | | | | | 3607 | 3207 | Basen kodierende Sequenz gesamt |
| RAF1 | chr3 | NM_002880.3 | RAF1 Exon 17 | 12.626.113 | 12.626.133 | 610 - 616 | 21 | | COSM97875: c.1837C>G (L613V) |
| | | | RAF1 Exon 7 | 12.645.689 | 12.645.709 | 254 - 260 | 21 | | COSM181063: c.770C>T (S257L) |
| | | | | | | | 42 | 42 | Basen kodierende Sequenz gesamt |
| RET | chr10 | NM_020975.5 | RET Exon 10 | 43.609.061 | 43.609.082 | 606 - 613 | 22 | | COSM95656: c.1827C>A (C609*) |
| | | | RET Exon 10 | 43.609.086 | 43.609.114 | 615 - 623 | 29 | | COSM29804: c.1858T>C (C620R) |
| | | | RET Exon 11 | 43.609.926 | 43.609.961 | 627 - 638 | 36 | 2 | COSM966: c.1900T>C (C634R) |
| | | | RET Exon 13 | 43.613.830 | 43.613.851 | 765 - 772 | 22 | | COSM4418405: c.2307G>T (L796L) |

| Gen | Lage (hg19) | Transkript | Amplifikat (Covered target region) | | | | | | Wichtige Mutationen (nach COSMIC) | |
|-------------|----------------|-------------|------------------------------------|--------------|------------|-------------|------------------------|---------------------------------------|---|--|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | Größe nicht- kodierende Sequenz | | |
| | | | RET Exon 13 | 43.613.895 | 43.613.919 | 787 - 794 | 25 | | COSM1159820: c.2372A>T (Y791F) | |
| | | | RET Exon 15 | 43.615.558 | 43.615.579 | 880 - 886 | 22 | | COSM1570338: c. 2651A>T (E884V) | |
| | | | RET Exon 15 | 43.615.583 | 43.615.604 | 888 - 894 | 22 | | COSM918124: c.2672C>T (S891L) | |
| | | | RET Exon 16 | 43.617.406 | 43.617.427 | 915 - 921 | 22 | | COSM965: c.2753T>C (M918T) | |
| | | | | | | | 200 | 198 | Basen kodierende Sequenz gesamt | |
| TP53 | chr17 | NM_000546.5 | TP53 Exon 11* | 7.572.917 | 7.573.018 | 367 - 394* | 102 | 20 | COSM13747: c.1146delA (K382fs) | |
| | | | TP53 Exon 10* | 7.573.917 | 7.574.043 | 332 - 367 | 127 | 20 | COSM11073: c.1024C>T (R342*) | |
| | | | NM_001276699 | TP53 Exon 6* | 7.576.527 | 7.576.594 | 173 - 188* | 68 | 20 | |
| | | | NM_001126116 | TP53 Exon 6* | 7.576.615 | 7.576.667 | 200 - 210* | 53 | 20 | |
| | | | TP53 Exon 9* | 7.576.843 | 7.576.936 | 307 - 331 | 94 | 20 | COSM10786: c.949C>T (Q317*) | |
| | | | TP53 Exon 8* | 7.577.009 | 7.577.165 | 261 - 307 | 157 | 20 | COSM10660: c.818G>A (R273H) | |
| | | | TP53 Exon 7* | 7.577.489 | 7.577.618 | 225 - 261 | 130 | 20 | COSM10662: c.743G>A (R248Q); COSM10656: c.742C>T (R248W) | |
| | | | TP53 Exon 6* | 7.578.167 | 7.578.299 | 187 - 224 | 133 | 20 | COSM10654: c.637C>T (R213*); COSM10758: c.659A>G (Y220C) | |
| | | | TP53 Exon 5* | 7.578.361 | 7.578.564 | 126 - 187 | 204 | 20 | COSM10648: c.524G>A (R175H); COSM10670: c.469G>T (V157F) | |
| | | | TP53 Exon 4* | 7.579.302 | 7.579.600 | 33 - 125 | 299 | 20 | COSM250061: c.215C>G (P72R); COSM44492: c.273G>A (W91*) | |
| | | | TP53 Exon 3* | 7.579.690 | 7.579.731 | 25 - 32 | 42 | 20 | COSM85573: c.80delC (P27fs) | |



| Gen | Lage (hg19) | Transkript | Amplifikat (Covered target region) | | | | | Wichtige Mutationen (nach COSMIC) | |
|-----|----------------|------------|------------------------------------|-----------|-----------|-------------|------------------------|--------------------------------------|---------------------------------------|
| | | | Name | Start | Stopp | Aminosäuren | Größe target region | | Größe nicht- kodierende Sequenz |
| | | | TP53 Exon 2* | 7.579.829 | 7.579.922 | 1 - 25 | 94 | 20 | COSM11606: c.31G>C (E11Q) |
| | | | | | | | 1503 | 1263 | Basen kodierende Sequenz gesamt |

Erklärung: vollständig untersuchte Exone sind mit * markiert, fett hervorgehoben wichtige Hotspotmutationen