

## Mutations in HCT 116

| HCT116_LARGE_INTESTINE      |                 |     |           |           |            |                        |              |                  |                |            |                       |                    |                      |
|-----------------------------|-----------------|-----|-----------|-----------|------------|------------------------|--------------|------------------|----------------|------------|-----------------------|--------------------|----------------------|
| <<   <   1   2   4   >   >> |                 |     |           |           |            |                        |              |                  |                |            |                       |                    |                      |
| Gene                        | Protein Change  | Chr | Start     | End       | NCBI Build | Variant Classification | Variant Type | Reference Allele | Tumor Allele 1 | dbSNP ID   | Annotation Transcript | Other Transcripts  | Mutation Assessor    |
| <a href="#">ABI1</a>        |                 | 10  | 27037488  | 27037488  | 37         | 3'UTR                  | Del          | A                | -              | rs75446996 | uc001isx.2            | ABI1_uc001ite.2... |                      |
| <a href="#">ACACA</a>       | p.V1545I        | 17  | 35545360  | 35545360  | 37         | Missense_Mutation      | SNP          | C                | T              |            | uc002hno.2            | ACACA_uc002hnk.... | <a href="#">view</a> |
| <a href="#">ACACA</a>       | p.V821A         | 17  | 35602129  | 35602129  | 37         | Missense_Mutation      | SNP          | A                | G              |            | uc002hno.2            | ACACA_uc002hnk.... | <a href="#">view</a> |
| <a href="#">ACACA</a>       | p.R682W         | 17  | 35608968  | 35608968  | 37         | Missense_Mutation      | SNP          | G                | A              |            | uc002hno.2            | ACACA_uc002hnk.... | <a href="#">view</a> |
| <a href="#">ACACA</a>       |                 | 17  | 35766564  | 35766564  | 37         | 5'UTR                  | Del          | A                | -              |            | uc002hno.2            | ACACA_uc002hnn.... |                      |
| <a href="#">ACVR2A</a>      | p.K435fs        | 2   | 148683686 | 148683686 | 37         | Frame_Shift_Del        | Del          | A                | -              |            | uc002twg.2            | ACVR2A_uc010zbn... |                      |
| <a href="#">ADAM10</a>      |                 | 15  | 58889719  | 58889719  | 37         | 3'UTR                  | SNP          | T                | C              |            | uc002afd.1            | ADAM10_uc010bgc... | <a href="#">view</a> |
| <a href="#">ADAM17</a>      |                 | 2   | 9630245   | 9630245   | 37         | 3'UTR                  | Del          | T                | -              |            | uc002qzu.2            | IAH1_uc010yiz.1... |                      |
| <a href="#">ADAM18</a>      | p.V554A         | 8   | 39537585  | 39537585  | 37         | Missense_Mutation      | SNP          | T                | C              |            | uc003xni.2            | ADAM18_uc010lww... | <a href="#">view</a> |
| <a href="#">ADAM28</a>      |                 | 8   | 24211994  | 24211997  | 37         | 3'UTR                  | Del          | CTAT             | -              | rs10610134 | uc003xdy.2            | ADAM28_uc011laa... |                      |
| <a href="#">ADAM28</a>      | p.L72fs         | 8   | 24167472  | 24167473  | 37         | Frame_Shift_Ins        | Ins          | -                | A              |            | uc003xdy.2            | ADAM28_uc003xdx... |                      |
| <a href="#">ADAM29</a>      | p.L691del       | 4   | 175898744 | 175898746 | 37         | In_Frame_Del           | Del          | TTA              | -              |            | uc003iuc.2            | ADAM29_uc003iud... |                      |
| <a href="#">ADAMTS19</a>    | p.Y1199C        | 5   | 129072883 | 129072883 | 37         | Missense_Mutation      | SNP          | A                | G              |            | uc003kvb.1            | ADAMTS19_uc010j... | <a href="#">view</a> |
| <a href="#">AFF3</a>        | p.I683N         | 2   | 100210150 | 100210150 | 37         | Missense_Mutation      | SNP          | A                | T              |            | uc002taf.2            | AFF3_uc002tag.2... | <a href="#">view</a> |
| <a href="#">AFF4</a>        | p.K924fs        | 5   | 132223814 | 132223814 | 37         | Frame_Shift_Del        | Del          | T                | -              |            | uc003kyd.2            | AFF4_uc011cxk.1... |                      |
| <a href="#">AKAP12</a>      | p.1531_1532insE | 6   | 151674116 | 151674117 | 37         | In_Frame_Ins           | Ins          | -                | GAG            | rs34737819 | uc011eep.1            | AKAP12_uc003qoe... |                      |
| <a href="#">AKAP4</a>       | p.M826V         | X   | 49955692  | 49955692  | 37         | Missense_Mutation      | SNP          | T                | C              |            | uc004dow.1            | AKAP4_uc004dov.... | <a href="#">view</a> |
| <a href="#">AKAP9</a>       | p.1335_1336insQ | 7   | 91652178  | 91652179  | 37         | In_Frame_Ins           | Ins          | -                | AAC            | rs34756483 | uc003ulg.2            | AKAP9_uc003ule.... |                      |
| <a href="#">AKAP9</a>       |                 | 7   | 91738049  | 91738049  | 37         | Intron                 | SNP          | C                | T              |            | uc003ulg.2            | AKAP9_uc003ulf.... | <a href="#">view</a> |
| <a href="#">ALPK1</a>       | p.F937fs        | 4   | 113353512 | 113353512 | 37         | Frame_Shift_Del        | Del          | T                | -              |            | uc003iap.3            | ALPK1_uc003ian.... |                      |
| <a href="#">ALS2</a>        |                 | 2   | 202587707 | 202587707 | 37         | Intron                 | Del          | A                | -              |            | uc002uyo.2            | ALS2_uc002uyp.3... |                      |
| <a href="#">ALS2</a>        | p.S694N         | 2   | 202609070 | 202609070 | 37         | Missense_Mutation      | SNP          | C                | T              |            | uc002uyo.2            | ALS2_uc002uyp.3... | <a href="#">view</a> |
| <a href="#">ARFGEF1</a>     | p.I1169fs       | 8   | 68140284  | 68140284  | 37         | Frame_Shift_Del        | Del          | T                | -              |            | uc003xxo.1            | ARFGEF1_uc003xx... |                      |
| <a href="#">ARFGEF1</a>     | p.R247G         | 8   | 68204259  | 68204259  | 37         | Missense_Mutation      | SNP          | T                | C              |            | uc003xxo.1            |                    | <a href="#">view</a> |
| <a href="#">ARHGAP29</a>    |                 | 1   | 94643144  | 94643144  | 37         | Intron                 | Del          | T                | -              |            | uc001dqj.3            | ARHGAP29_uc009w... |                      |
| <a href="#">ASPH</a>        |                 | 8   | 62602296  | 62602296  | 37         | Intron                 | Del          | A                | -              |            | uc003xuj.2            | ASPH_uc011leg.1... |                      |
| <a href="#">ASPSCR1</a>     | p.A240fs        | 17  | 79954508  | 79954508  | 37         | Frame_Shift_Del        | Del          | C                | -              |            | uc002kcy.2            | ASPSCR1_uc002kc... |                      |
| <a href="#">ATIC</a>        | p.W527*         | 2   | 216213894 | 216213894 | 37         | Nonsense_Mutation      | SNP          | G                | A              |            | uc002vex.3            | ATIC_uc010zjo.1... | <a href="#">view</a> |
| <a href="#">ATP11A</a>      | p.T367M         | 13  | 113481084 | 113481084 | 37         | Missense_Mutation      | SNP          | C                | T              |            | uc001vsj.3            | ATP11A_uc001vsi... | <a href="#">view</a> |
| <a href="#">ATRX</a>        | p.T1529del      | X   | 76891517  | 76891519  | 37         | In_Frame_Del           | Del          | TGG              | -              |            | uc004ecp.3            | ATRX_uc004ecq.3... |                      |
| <a href="#">AXL</a>         | p.M589K         | 19  | 41758310  | 41758310  | 37         | Missense_Mutation      | SNP          | T                | A              |            | uc010ehj.2            | CYP2F1_uc010xvw... | <a href="#">view</a> |
| <a href="#">BAI1</a>        | p.P438fs        | 8   | 143558835 | 143558835 | 37         | Frame_Shift_Del        | Del          | C                | -              |            | uc003ywm.2            |                    |                      |
| <a href="#">BAI1</a>        | p.G1321S        | 8   | 143623556 | 143623556 | 37         | Missense_Mutation      | SNP          | G                | A              |            | uc003ywm.2            |                    | <a href="#">view</a> |
| <a href="#">BAX</a>         | p.M38fs         | 19  | 49458971  | 49458971  | 37         | Frame_Shift_Del        | Del          | G                | -              |            | uc002plf.1            | BAX_uc002plg.1...  |                      |
| <a href="#">BCKDK</a>       | p.R131C         | 16  | 31121413  | 31121413  | 37         | Missense_Mutation      | SNP          | C                | T              |            | uc002eaw.3            | BCKDK_uc002eav.... | <a href="#">view</a> |
| <a href="#">BCL11A</a>      | p.S763fs        | 2   | 60687759  | 60687760  | 37         | Frame_Shift_Ins        | Ins          | -                | T              |            | uc002sae.1            | BCL11A_uc002sab... |                      |
| <a href="#">BCL11A</a>      | p.R258C         | 2   | 60689275  | 60689275  | 37         | Missense_Mutation      | SNP          | G                | A              |            | uc002sae.1            | BCL11A_uc002sab... | <a href="#">view</a> |
| <a href="#">BCL9</a>        | p.A242T         | 1   | 147090685 | 147090685 | 37         | Missense_Mutation      | SNP          | G                | A              |            | uc001epq.2            | BCL9_uc010ozr.1... | <a href="#">view</a> |

|                          |            |    |           |           |    |                   |     |     |    |                      |            |                    |                      |
|--------------------------|------------|----|-----------|-----------|----|-------------------|-----|-----|----|----------------------|------------|--------------------|----------------------|
| <a href="#">BMPR2</a>    | p.E581fs   | 2  | 203420130 | 203420130 | 37 | Frame_Shift_Del   | Del | A   | -  |                      | uc002uzf.3 | BMPR2_uc010ftr.... |                      |
| <a href="#">BRCA2</a>    | p.I2672fs  | 13 | 32937354  | 32937355  | 37 | Frame_Shift_Ins   | Ins | -   | A  | rs116677141          | uc001uub.1 |                    |                      |
| <a href="#">BRD3</a>     | p.P24fs    | 9  | 136918529 | 136918529 | 37 | Frame_Shift_Del   | Del | G   | -  |                      | uc004cew.2 | BRD3_uc004cex.2... |                      |
| <a href="#">BRSK1</a>    | p.D392fs   | 19 | 55815036  | 55815036  | 37 | Frame_Shift_Del   | Del | C   | -  |                      | uc002qkf.2 | BRSK1_uc002qkg.... |                      |
| <a href="#">C15ORF2</a>  | p.M389I    | 15 | 24922181  | 24922181  | 37 | Missense_Mutation | SNP | G   | T  |                      | uc001ywo.2 |                    | <a href="#">view</a> |
| <a href="#">C15ORF55</a> | p.R243S    | 15 | 34640796  | 34640796  | 37 | Missense_Mutation | SNP | C   | A  |                      | uc010ucc.1 | C15orf55_uc010u... | <a href="#">view</a> |
| <a href="#">C9ORF96</a>  | p.R191C    | 9  | 136256560 | 136256560 | 37 | Missense_Mutation | SNP | C   | T  |                      | uc004cdk.2 | C9orf96_uc004cd... | <a href="#">view</a> |
| <a href="#">CACNA1F</a>  | p.D1760Y   | X  | 49063303  | 49063303  | 37 | Missense_Mutation | SNP | C   | A  |                      | uc004dmb.2 | CACNA1F_uc010ni... | <a href="#">view</a> |
| <a href="#">CACNB2</a>   |            | 10 | 18828721  | 18828722  | 37 | 3'UTR             | Ins | -   | AA | rs3841459            | uc001ipr.2 | CACNB2_uc009xjz... |                      |
| <a href="#">CASP1</a>    |            | 11 | 104896928 | 104896928 | 37 | Intron            | SNP | C   | T  |                      | uc010rve.1 | CASP1_uc001pig...  | <a href="#">view</a> |
| <a href="#">CASP2</a>    | p.A398V    | 7  | 143001842 | 143001842 | 37 | Missense_Mutation | SNP | C   | T  |                      | uc003wco.2 | CASP2_uc003wcp...  | <a href="#">view</a> |
| <a href="#">CASP4</a>    |            | 11 | 104822590 | 104822590 | 37 | Intron            | Del | T   | -  |                      | uc001pid.1 | CASP4_uc001pib...  |                      |
| <a href="#">CCR1</a>     | p.N67fs    | 3  | 46245605  | 46245605  | 37 | Frame_Shift_Del   | Del | T   | -  |                      | uc003cph.1 | CCR3_uc003cpg.1... |                      |
| <a href="#">CCR6</a>     |            | 6  | 167416821 | 167416821 | 37 | 5'UTR             | Del | T   | -  |                      | uc003qvl.2 | FGFR1OP_uc011eg... |                      |
| <a href="#">CDH1</a>     | p.V119fs   | 16 | 68835766  | 68835766  | 37 | Frame_Shift_Del   | Del | G   | -  |                      | uc002ewg.1 | CDH1_uc010vlj.1... |                      |
| <a href="#">CDK12</a>    | p.P250H    | 17 | 37619073  | 37619073  | 37 | Missense_Mutation | SNP | C   | A  |                      | uc010cvv.2 | CDK12_uc010wef...  | <a href="#">view</a> |
| <a href="#">CDK14</a>    | p.I321V    | 7  | 90613476  | 90613476  | 37 | Missense_Mutation | SNP | A   | G  |                      | uc003uky.2 | CDK14_uc003ukz.... | <a href="#">view</a> |
| <a href="#">CDK8</a>     | p.V195A    | 13 | 26959417  | 26959417  | 37 | Missense_Mutation | SNP | T   | C  |                      | uc001uqr.1 | CDK8_uc001uqs.1... | <a href="#">view</a> |
| <a href="#">CDKL2</a>    | p.K316fs   | 4  | 76523333  | 76523333  | 37 | Frame_Shift_Del   | Del | T   | -  |                      | uc011cbp.1 | CDKL2_uc003hiq.... |                      |
| <a href="#">CDX2</a>     | p.V306fs   | 13 | 28537278  | 28537278  | 37 | Frame_Shift_Del   | Del | C   | -  |                      | uc001urv.2 |                    |                      |
| <a href="#">CEP110</a>   | p.T1383I   | 9  | 123914947 | 123914947 | 37 | Missense_Mutation | SNP | C   | T  |                      | uc004bkk.1 | CEP110_uc004bla... | <a href="#">view</a> |
| <a href="#">CEP110</a>   |            | 9  | 123939699 | 123939699 | 37 | 3'UTR             | Del | T   | -  | rs75910820           | uc004bkk.1 | CEP110_uc004blb... |                      |
| <a href="#">CHD1</a>     | p.P1684del | 5  | 98192165  | 98192167  | 37 | In_Frame_Del      | Del | AGG | -  | s79267787;rs61749614 | uc003knf.2 | CHD1_uc010jbn.2... |                      |
| <a href="#">CHD1</a>     | p.S126F    | 5  | 98238664  | 98238664  | 37 | Missense_Mutation | SNP | G   | A  |                      | uc003knf.2 |                    | <a href="#">view</a> |
| <a href="#">CHD1</a>     | p.A95V     | 5  | 98239584  | 98239584  | 37 | Missense_Mutation | SNP | G   | A  |                      | uc003knf.2 |                    | <a href="#">view</a> |
| <a href="#">CHD8</a>     | p.R1964C   | 14 | 21860710  | 21860710  | 37 | Missense_Mutation | SNP | G   | A  |                      | uc001was.1 | CHD8_uc001war.1... | <a href="#">view</a> |
| <a href="#">CHD8</a>     | p.R1576C   | 14 | 21862472  | 21862472  | 37 | Missense_Mutation | SNP | G   | A  |                      | uc001was.1 | CHD8_uc001war.1... | <a href="#">view</a> |
| <a href="#">CHD8</a>     | p.G796W    | 14 | 21873452  | 21873452  | 37 | Missense_Mutation | SNP | C   | A  |                      | uc001was.1 | CHD8_uc001war.1... | <a href="#">view</a> |
| <a href="#">CHD9</a>     |            | 16 | 53358883  | 53358883  | 37 | 3'UTR             | SNP | T   | C  |                      | uc002ehb.2 | CHD9_uc002egy.2... | <a href="#">view</a> |
| <a href="#">CHEK2</a>    | p.L398P    | 22 | 29092920  | 29092920  | 37 | Missense_Mutation | SNP | A   | G  |                      | uc003adt.1 | CHEK2_uc003ads.... | <a href="#">view</a> |
| <a href="#">CNTN5</a>    | p.V442A    | 11 | 99942462  | 99942462  | 37 | Missense_Mutation | SNP | T   | C  |                      | uc001pga.2 | CNTN5_uc009yvw.... | <a href="#">view</a> |
| <a href="#">CNTN6</a>    |            | 3  | 1445212   | 1445212   | 37 | 3'UTR             | Del | A   | -  | rs74788015           | uc003boz.2 | CNTN6_uc011asj.... |                      |
| <a href="#">COL1A1</a>   | p.R1141*   | 17 | 48264847  | 48264847  | 37 | Nonsense_Mutation | SNP | G   | A  | rs72656314           | uc002iqm.2 |                    | <a href="#">view</a> |
| <a href="#">COL3A1</a>   | p.E682D    | 2  | 189864034 | 189864034 | 37 | Missense_Mutation | SNP | A   | C  |                      | uc002uqj.1 |                    | <a href="#">view</a> |
| <a href="#">CPNE5</a>    | p.D38D     | 6  | 36790856  | 36790856  | 37 | Silent            | SNP | G   | A  |                      | uc003omr.1 | CPNE5_uc003oms.... | <a href="#">view</a> |
| <a href="#">CSMD3</a>    |            | 8  | 114389107 | 114389107 | 37 | Intron            | SNP | G   | A  |                      | uc003ynu.2 | CSMD3_uc003ynt.... | <a href="#">view</a> |
| <a href="#">CTBP2</a>    | p.G140R    | 10 | 126715911 | 126715911 | 37 | Missense_Mutation | SNP | C   | T  |                      | uc001lie.3 | CTBP2_uc009yak.... | <a href="#">view</a> |
| <a href="#">CTNNB1</a>   | p.S45del   | 3  | 41266134  | 41266136  | 37 | In_Frame_Del      | Del | CTT | -  |                      | uc010hia.1 | CTNNB1_uc003ckp... |                      |
| <a href="#">CXCL12</a>   |            | 10 | 44873992  | 44873993  | 37 | Intron            | Ins | -   | CT | rs117617571          | uc001jhb.2 | CXCL12_uc001jfb... |                      |
| <a href="#">CXCR4</a>    | p.Y80C     | 2  | 136873271 | 136873271 | 37 | Missense_Mutation | SNP | T   | C  |                      | uc002tuy.2 | CXCR4_uc010fnk.... | <a href="#">view</a> |
| <a href="#">CYP27B1</a>  | p.E63D     | 12 | 58160636  | 58160636  | 37 | Missense_Mutation | SNP | C   | A  |                      | uc001spz.1 | CYP27B1_uc001sq... | <a href="#">view</a> |
| <a href="#">DAB1</a>     | p.V193I    | 1  | 57536716  | 57536716  | 37 | Missense_Mutation | SNP | C   | T  |                      | uc001cys.1 | DAB1_uc001cyt.1... | <a href="#">view</a> |
| <a href="#">DACH1</a>    |            | 13 | 72014626  | 72014628  | 37 | 3'UTR             | Del | TTT | -  |                      | uc010thn.1 | DACH1_uc010tho.... |                      |

|                          |           |    |           |           |    |                   |     |   |   |             |            |                    |                      |
|--------------------------|-----------|----|-----------|-----------|----|-------------------|-----|---|---|-------------|------------|--------------------|----------------------|
| <a href="#">DACH1</a>    | p.T596fs  | 13 | 72053384  | 72053385  | 37 | Frame_Shift_Ins   | Ins | - | T |             | uc010thn.1 | DACH1_uc010tho.... |                      |
| <a href="#">DAPK2</a>    | p.R296C   | 15 | 64204369  | 64204369  | 37 | Missense_Mutation | SNP | G | A |             | uc002amr.2 | DAPK2_uc010uim.... | <a href="#">view</a> |
| <a href="#">DDIT3</a>    |           | 12 | 57910472  | 57910472  | 37 | 3'UTR             | SNP | T | C |             | uc009zps.2 | MARS_uc001sof.1... | <a href="#">view</a> |
| <a href="#">DDR2</a>     | p.M117I   | 1  | 162724579 | 162724579 | 37 | Missense_Mutation | SNP | G | A |             | uc001gcf.2 | DDR2_uc001gcg.2... | <a href="#">view</a> |
| <a href="#">DICER1</a>   | p.V288I   | 14 | 95592958  | 95592958  | 37 | Missense_Mutation | SNP | C | T |             | uc001ydw.2 | DICER1_uc001ydv... | <a href="#">view</a> |
| <a href="#">DLC1</a>     | p.V1025A  | 8  | 12956001  | 12956001  | 37 | Missense_Mutation | SNP | A | G |             | uc003wwm.2 | DLC1_uc003wwk.1... | <a href="#">view</a> |
| <a href="#">DOCK3</a>    | p.T1850fs | 3  | 51417604  | 51417604  | 37 | Frame_Shift_Del   | Del | C | - |             | uc011bds.1 |                    |                      |
| <a href="#">DST</a>      | p.E2927K  | 6  | 56420143  | 56420143  | 37 | Missense_Mutation | SNP | C | T |             | uc003pdf.2 | DST_uc003pcz.3_... | <a href="#">view</a> |
| <a href="#">DST</a>      |           | 6  | 56485020  | 56485020  | 37 | Intron            | Del | T | - |             | uc003pdf.2 | DST_uc003pcz.3_... |                      |
| <a href="#">DSTYK</a>    | p.V666A   | 1  | 205129350 | 205129350 | 37 | Missense_Mutation | SNP | A | G |             | uc001hbw.2 | DSTYK_uc001hbx.... | <a href="#">view</a> |
| <a href="#">DSTYK</a>    | p.G661fs  | 1  | 205129365 | 205129365 | 37 | Frame_Shift_Del   | Del | C | - |             | uc001hbw.2 | DSTYK_uc001hbx.... |                      |
| <a href="#">EEF1D</a>    | p.K648N   | 8  | 144662293 | 144662293 | 37 | Missense_Mutation | SNP | C | A |             | uc003yyq.1 | NAPRT1_uc003yym... | <a href="#">view</a> |
| <a href="#">EIF4E</a>    | p.N84K    | 4  | 99809073  | 99809073  | 37 | Missense_Mutation | SNP | A | C |             | uc011ceb.1 | EIF4E_uc011cea.... | <a href="#">view</a> |
| <a href="#">EIF4EBP1</a> | p.A107V   | 8  | 37914773  | 37914773  | 37 | Missense_Mutation | SNP | C | T |             | uc003xks.2 |                    | <a href="#">view</a> |
| <a href="#">EP300</a>    | p.K1468fs | 22 | 41566525  | 41566525  | 37 | Frame_Shift_Del   | Del | A | - |             | uc003azl.3 |                    |                      |
| <a href="#">EPHA5</a>    |           | 4  | 66189753  | 66189754  | 37 | 3'UTR             | Ins | - | T | rs67487372  | uc011cah.1 | EPHA5_uc003hcx.... |                      |
| <a href="#">EPHA5</a>    | p.R701fs  | 4  | 66230873  | 66230873  | 37 | Frame_Shift_Del   | Del | T | - |             | uc011cah.1 | EPHA5_uc003hcx.... |                      |
| <a href="#">EPHA6</a>    |           | 3  | 97367230  | 97367231  | 37 | Intron            | Ins | - | A | rs115253553 | uc010how.1 | EPHA6_uc003drs.... |                      |
| <a href="#">ERBB3</a>    | p.Q261*   | 12 | 56481853  | 56481853  | 37 | Nonsense_Mutation | SNP | C | T |             | uc001sjh.2 | ERBB3_uc009zpj.... | <a href="#">view</a> |

Method: Hybrid Capture

