

The Many Roads from Alternative Splicing to Cancer. Molecular Mechanisms Involving Driver Genes

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SUPPLEMENTARY MATERIAL

Supplementary Table S1. Cancer driver genes regulated by alternative splicing¹

Gene	Splicing events ¹	Mechanism ²	References
<i>TP53</i>	esk, IR, at, ai, ce, me	mcis	[96,191,192]
<i>KRAS</i>	esk, at, a5', ce, me	mcis, ep	[181,182,193–195]
<i>BRAF</i>	esk, a5'	N/R	[196,197]
<i>ANAPC1</i>	esk	mcis	[198]
<i>ARID1A</i>	a3'	ocis	[109,110]
<i>KMT2C</i>	esk, a3'	SF	[120]
<i>PTEN</i>	esk, IR, at	mcis	[97,99,101,192]
<i>NRAS</i>	esk, at, o	N/R	[199,200]
<i>CTNNB1</i>	esk	SF	[121]
<i>CDKN2A</i>	esk, at, ai, a5'	mcis	[104,201–204]
<i>IDH1</i>	IR, at	mcis	[10,205]
<i>RB1</i>	esk	mcis	[206,207]
<i>ATM</i>	esk	mcis	[208]
<i>NF1</i>	esk, ce	mcis	[209]
<i>SMAD4</i>	esk, ai	mcis	[210]
<i>FAT1</i>	IR, ai	N/R	[211]
<i>FBXW7</i>	ai	N/R	[212]
<i>NOTCH1</i>	at	mcis	[213]
<i>EGFR</i>	esk, at	mcis, SF, ep	[122,214–218]
<i>KDM6A</i>	a3'	esk	[219]
<i>ATRX</i>	esk	mcis, o	[220–222]
<i>SETD2</i>	o	mcis	[223]
<i>ERBB2</i>	esk	mcis	[224]
<i>NFE2L2</i>	esk, ai	mcis, o	[34]
<i>CDH1</i>	a3'	mcis, SF	[123]
<i>VHL</i>	esk, ai	SF	[225–227]
<i>KEAP1</i>	esk, ai	N/R	[228,229]
<i>EP300</i>	esk	SF	[124]
<i>PIK3R1</i>	esk	mcis	[201,219]
<i>GATA3</i>	a3'	mcis	[201,221]
<i>AR</i>	at, a3', ce	SF, ep	[179,230–232]
<i>FGFR3</i>	esk, me	N/R	[233–235]
<i>HRAS</i>	esk, a3', a5'	mcis	[181,200,236–238]

<i>BRCA2</i>	esk	mcis	[239–241]
<i>BAP1</i>	a3'	mcis	[221,223]
<i>AKT1</i>	ce	N/R	[242,243]
<i>STK11</i>	esk, at, a3'	mcis	[244,245]
<i>ESR1</i>	esk, at, ai, a3', ce	mcis, ocis	[108,246–249]
<i>PTCH1</i>	esk, ai, ce	mcis, SF	[114,250,251]
<i>NOTCH2</i>	esk, at	mcis	[252,253]
<i>CTCF</i>	esk, ai	N/R	[254]
<i>CASP8</i>	a3'	N/R	[255]
<i>CDK12</i>	a5'	mcis	[256]
<i>KIT</i>	ai, o	mcis	[257–259]
<i>HLA-A</i>	IR	N/R	[260,261]
<i>MYD88</i>	esk, IR, ai, a5'	SF	[125–127]
<i>TCF7L2</i>	esk, a5'	SF	[128,262,263]
<i>AXIN1</i>	esk	N/R	[264]
<i>PPP2R1A</i>	esk, a3', a5'	N/R	[265]
<i>FGFR2</i>	me	SF, ep	[70,79,180,266–268]
<i>ELF3</i>	esk, a5'	mcis	[269]
<i>DNMT3A</i>	esk, at, ai, ce	ep	[270–275]
<i>BCL2</i>	at	N/R	[276]
<i>EZH2</i>	esk, ai, a5', ce	mcis, SF	[79,104,111–113,140,277–280]
<i>ALK</i>	esk, ai	ep	[176,281,282]
<i>IDH2</i>	IR	mcis	[10,283]
<i>RHOA</i>	esk	SF	[284]
<i>F3B1</i>	esk	SF	[285]
<i>MED12</i>	esk	mcis	[286]
<i>TGFBR2</i>	esk	mcis	[287,288]
<i>RUNX1</i>	ai	N/R	[289–291]
<i>MAX</i>	me	SF	[131]
<i>IRF4</i>	esk, at, ai, a3', a5'	SF	[292]
<i>KDR</i>	at	N/R	[293]
<i>SGK1</i>	IR, a3'	mcis	[294]
<i>PRDM1</i>	ai	ep	[167]
<i>CCND1</i>	at	mcis	[295]
<i>NF2</i>	esk	N/R	[296]
<i>MYCN</i>	esk	o	[297,298]
<i>FLT3</i>	esk, a3', a5'	N/R	[252]
<i>RAC1</i>	ce	SF	[133,299]
<i>BRCA1</i>	esk	mcis	[300–302]
<i>RET</i>	at	N/R	[303,304]
<i>MEN1</i>	IR	mcis	[305]
<i>CACNA1D</i>	esk	N/R	[306]
<i>SMAD3</i>	a5'	o	[307]
<i>BCLAF1</i>	ce	SF	[134]
<i>STAT3</i>	a3', o	N/R	[308–310]
<i>ERBB4</i>	esk	N/R	[311,312]
<i>MET</i>	esk	N/R	[313–315]
<i>FN1</i>	esk	SF, o	[316–319]

<i>TET1</i>	at	ep	[175]
<i>IKZF1</i>	esk	N/R	[320,321]
<i>FANCD2</i>	at	SF	[135]
<i>EIF1AX</i>	IR, ce	mcis	[322]
<i>FGFR1</i>	esk, IR, ce	N/R	[235,323–325]
<i>TP63</i>	esk, ai, ce	ep	[105,106]
<i>RUNX1T1</i>	ce	N/R	[326]
<i>PTPRC</i>	esk	mcis	[261]
<i>BCR</i>	esk	N/R	[327]
<i>BCL6</i>	esk	N/R	[328]
<i>ABL2</i>	esk	N/R	[329]
<i>BTB</i>	esk, a3', a5'	N/R	[330]
<i>DICER1</i>	me	N/R	[331]
<i>PTPN14</i>	a5'	N/R	[332]
<i>EPHA7</i>	at	N/R	[333]
<i>DAXX</i>	a3', a5'	N/R	[334]
<i>PRF1</i>	sk	mcis	[104]
<i>POLE</i>	at	mcis	[335]
<i>PSIP1</i>	ce	N/R	[71,336]
<i>PRKCD</i>	a5'	SF	[136]
<i>JAK2</i>	esk	mcis	[48,337]
<i>ABCB1</i>	esk, ae	mcis	[338]
<i>ABL1</i>	esk, ce	mcis	[339]
<i>ETV6</i>	esk, at	mcis	[340]
<i>MAP2K7</i>	esk	SF	[137]
<i>IRF1</i>	esk, at,	mcis, SF	[138,341,342]
<i>TGIF1</i>	esk, ai	N/R	[343]
<i>TNC</i>	ce	N/R	[344]
<i>CASZ1</i>	esk	N/R	[345]
<i>NSD2</i>	esk, ai	N/R	[40]
<i>SALL4</i>	a5'	N/R	[346]
<i>ETV5</i>	ai	N/R	[347]
<i>SMARCB1</i>	a5'	mcis	[348]
<i>FAS</i>	esk	N/R	[349]
<i>NTKR3</i>	at	o	[350]
<i>DHX9</i>	esk, ce	SF, o	[142,351]
<i>FOXO1</i>	o	mcis	[352]
<i>FOXP1</i>	esk, ai	mcis, ep	[170,353]
<i>CDK4</i>	esk, at	mcis	[104,354,355]
<i>DIS3</i>	me	mcis	[356,357]
<i>ERG</i>	esk	N/R	[358]
<i>NPM1</i>	esk	N/R	[359,360]
<i>SMC1A</i>	esk, at, ai	SF	[143]
<i>KLF4</i>	esk	SF	[144]
<i>ING1</i>	at, ai	N/R	[361]
<i>PTPN6</i>	esk, ai	N/R	[362]
<i>PRDM2</i>	ai	ep	[171,173]
<i>RBM39</i>	ce	SF	[363]

<i>CCND2</i>	esk, at	mcis, SF, o	[114,364]
<i>AKT3</i>	esk, ai	SF, ep	[122,168]
<i>NTRK1</i>	esk	o	[365–367]
<i>ARAF</i>	at	SF	[145,146]
<i>SUSD2</i>	IR	N/R	[368]
<i>RBFOX2</i>	esk, a3'	N/R	[369]
<i>CEBPA</i>	ai	N/R	[370]
<i>CIITA</i>	esk	N/R	[371]
<i>PRRX1</i>	me	N/R	[372,373]
<i>FGFR4</i>	IR	N/R	[374]
<i>RAP1GDS1</i>	esk	SF	[147,150–152]
<i>ZNF148</i>	ai, ce	N/R	[375,376]
<i>RELA</i>	esk, a5'	N/R	[377]
<i>NKX2-1</i>	ai	SF	[378,379]
<i>RBM38</i>	esk, at	N/R	[380]
<i>QKI</i>	at	N/R	[381]
<i>MDM4</i>	IR, at	SF	[121,133,382–385]
<i>CSF3R</i>	esk, ce	N/R	[386]
<i>GATA1</i>	esk, ai	mcis	[387]
<i>CDX2</i>	ai, a5'	N/R	[388,389]
<i>CHEK2</i>	esk, a3', a5'	mcis	[219,390,391]
<i>HGF</i>	at	N/R	[392,393]
<i>DAZAP1</i>	at	o	[394]
<i>NRP1</i>	esk, at	N/R	[395,396]
<i>MSI2</i>	ai	N/R	[397]
<i>XPC</i>	esk, a3'	mcis, o	[398]
<i>IKZF3</i>	esk	N/R	[399]
<i>BCL11A</i>	at, ce	N/R	[400,401]
<i>GLI1</i>	esk, ai	N/R	[402,403]
<i>MDM2</i>	esk, ai, a3'	N/R	[404]
<i>PML</i>	esk, at	N/R	[405–407]
<i>CBFA2T3</i>	esk, at	ep	[174]
<i>FANCA</i>	esk, ce	mcis	[408,409]
<i>NUMA1</i>	esk, at	SF	[53,410,411]
<i>AFF1</i>	a3'	N/R	[412,413]
<i>CXCR4</i>	IR, ai	N/R	[414,415]
<i>EWSR1</i>	IR	SF	[148]

¹ Abbreviations used: esk, exon skipping; IR, intron retention; at, alternative termination; ai, alternative initiation; a3' alternative 3' splice site; a5', alternative 5' splice site; ce, inclusion of cryptic exon; me, mutual exclusive exons; o, other events.

² Molecular mechanisms involved in the splicing events. Abbreviations used: mcis, mutation in *cis* elements; ocis, other causes involving *cis* elements; SF, changes in splicing factors (mutations, change of level); ep, changes in epigenetic marks or chromatin structure; o, other causes.

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