Supplemental Data 2. Splicing diagrams and nucleotide sequence of exon-exon RT-PCR products of genes showing abnormal splicing events around U12 introns in SCNM1-deficient cells.

For each gene, the sequence of RT-PCR products corresponding to normally, or alternatively (aberrant) spliced transcripts (designated with numbers), together with a schematic representation (not to scale) of splicing events is included. Exons are designated with boxes and introns with lines. U12 introns are specified with a red line and exons flanking U12 introns are in blue and green. In the case of *DERL2*, which has 2 contiguous U12 introns, only the one found as significantly retained in SCNM1-deficient fibroblasts through bioinformatics analysis is between blue and green exons. Dotted lines indicate splicing junctions and the nucleotide sequences of relevant 5' and 3' splice sites around U12 introns are indicated. The cDNA sequence of each transcript is shown below its corresponding diagram. Different code of letters was used: blue and black for canonical exons (capital letters), and red (lower case letters) for U12 intronic sequences incorporated into transcripts. Additionally, in the case of variant 2 of both, *FAM92A* and *RABL2B,* purple color was used to separate splicing events occurring within U12 introns. For *DERL2* and *ZC3H8*, nucleotides which are not present in some of the aberrant isoforms are underlined in the sequence of the normally spliced transcript.

Abbreviations: CU12IR (Complete U12 Intron Retention), A5SS (Activation of cryptic 5' Splice Site [within U12 intron or adjacent exons]), A3SS (Activation of cryptic 3' Splice Site [within U12 intron or adjacent exons]), ES (Exon Skipping) and CEI (Cryptic Exon Inclusion).

AHCTF1

>Normally spliced transcript (NM_015446.5). Nucleotide sequence extends from exon 16 to exon 21.

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ACCTATTAGACGGCGTTACTGAAGCAGCCAAACACTCTATTACCATTTATTTGCTACTTGATAT
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TCTTGGGGCCAAGTTAAACTTATTCAGGGGTTTTGGTTGATAGATCATAATGACTATGAGAGTG
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TCAGGCATTCATGAGTCAGGGCG

>1 (CU12IR)

GGGCTTTTACCAGAAGGCATAGATGATTCTGTGCAGTTGTCAAGGTTATGCTACAACTACCCTG
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GTTTCATCCAGCTACTGCAAAACCTTTGTCATGGCAACATTCAAAGATTATTCAGGCATTCATG
AGTCAGGGCG

C17orf75

>Normally spliced transcript (NM_022344.4). Nucleotide sequence extends from exon 3 to exon 8.

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AACCCTTCAGAATATGTGGTCTGTTTTTTAGGAGGGTCTGAAAAAGGACTTGAGCTTTTCAGGC
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GAGTCACATAAAATCCTATCTGAGCAGCTGGTTTGAGGATGTTGTATGCCCAATCCAAAGGGTT
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TCAAGGAC

>1 (CU12IR)

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AACCCTTCAGAATATGTGGTCTGTTTTTTAGGAGGGTCTGAAAAAGGACTTGAGCTatatcctt
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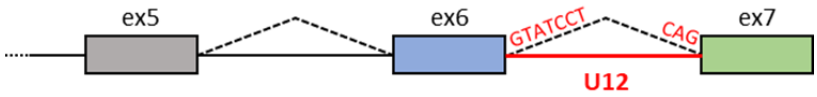
DCTN3

>Normally spliced transcript (NM_007234.5). Nucleotide sequence extends from exon 5 to exon 7.



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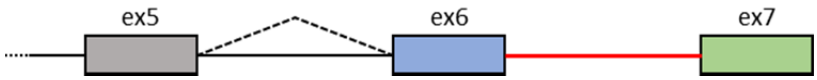
>1 (CU12IR)



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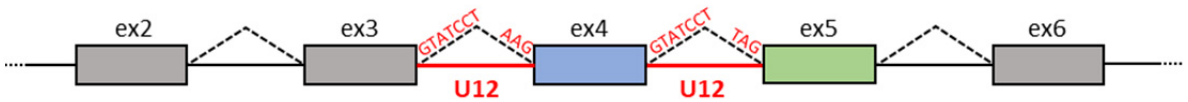
DERL2

>Normally spliced transcript (NM_016041.5). Nucleotide sequence extends from exon 1 to exon 6.



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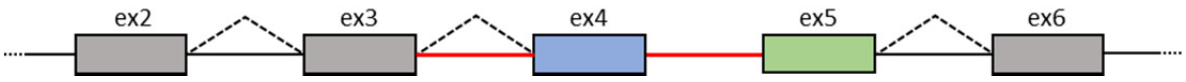
>1 (CU12IR)



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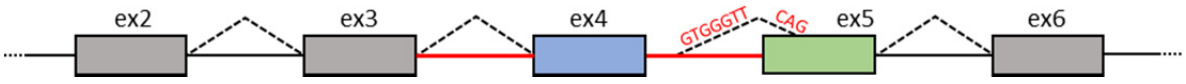
>2 (A5SS & A3SS)



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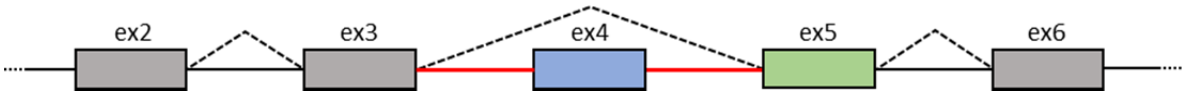
>3 (ES [Ex4])



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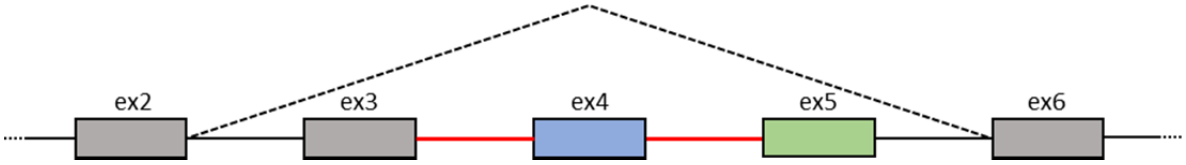
>4 (ES [Ex3, Ex4 & Ex5])



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DOCK1

>Normally spliced transcript (NM_001290223.2). Nucleotide sequence extends from exon 39 to exon 45.

GCCGAGCAGTATGAGAACGAAATGTTTGATTATGAGCAACTCAGCGAATTGCTGAAAAAACAGG
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CTACGGACAAGGGTTCCCCACATTCCTGCGGGGAAAAGTTTTCATTTACCGAGGGAAAGAGTAT
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G

>1 (CU12IR)

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GAGAATG

FAM92A (CIBAR1)

>Normally spliced transcript (NM_145269.5). Nucleotide sequence extends from exon 2 to exon 7.

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CACAGGCAGAAACGGAATTACAGAGAGCTGCAATGGATGCTAGCCGAACAAGTCGTCATCTGGA
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>1 (A5SS & ES [Ex5])

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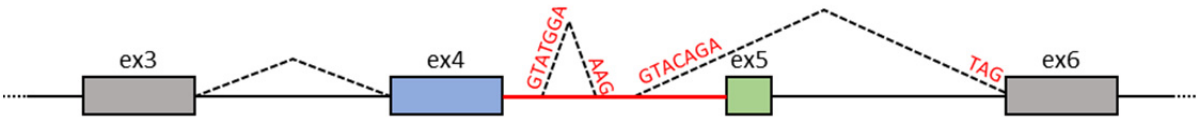
>2 (A5SS, CEI & ES [Ex5])



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>3 (A5SS & ES [Ex5])

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>4 (ES [Ex4 & Ex5])

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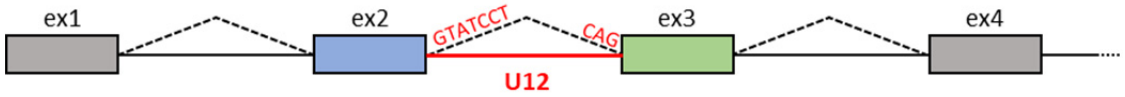
KRTCAP2

>Normally spliced transcript (NM_173852.4). Nucleotide sequence extends from exon 1 to exon 5.



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>1 (CU12IR)

TTCAGCTCGCCTTTCTTGGCCAGAGGCGCCGGTTGGACTCACGGGCGGGGCATGATGGTGGTGG
GTACGGGCACCTCGCTGGCGCTCTCCTCCCTCCTGTCCCTGCTGCTCTTTGCTGGGATGCAGAT
GTACAGCCGTCAGCTGGCCTCCACCGAGTGGCTCACCATCCAGGGCGGCCTGCTTGGTTCGGGT
CTCTTCGTGTTCTCGCTCACTgtatcctccctgcagttggaggggcgggccacgtaggcatgt
gcccttccccttccccacacagctctgtccccgttgcacaccctactccttaactccctcaacc
agGCCTTCAATAATCTGGAGAATCTTGTCTTTGGCAAAGGATTCCAAGCAAAGATCTTCCCTGA
GATTCTCCTGTGCCTCCTGTTGGCTCTCTTTGCATCTGGCCTCATCCACCGAGTCTGTGTCACC
ACCTGC

MARCHF6

>Normally spliced transcript (NM_005885.4). Nucleotide sequence extends from exon 7 to exon 16.

TAACCCACCAGCTGAGAACGCAGTGGTGGGGGAAAACCCTGATGCCCAGGATGACCAGGCAGAA
GAGGAGGAGGAGGACAATGAGGAGGAAGATGACGCTGGTGTGGAGGATGCGGCAGATGCTAATA
ACGGAGCCCAGGATGACATGAATTGGAATGCTTTAGAATGGGACCGAGCTGCTGAAGAGCTTAC
ATGGGAAAGAATGCTAGGACTTGATGGATCACTAGTTTTTCTGGAACATGTCTTCTGGGTGGTA

TCTTTAAATACACTGTTCATTCTTGTTTTTGCATTTTGCCCTTACCATATTGGTCATTTCTCCC
TTGTTGGTTTGGGATTTGAAGAACACGTCCAAGCATCTCATTTTGAAGGCCTAATCACAACCAT
AGTTGGGTATATACTTTTAGCAATAACACTGATAATTTGTCATGGCTTGGCAACTCTTGTGAAA
TTTCATAGATCTCGTCGCTTACTGGGAGTCTGCTATATTGTTGTTAAGGTCTCTTTGTTAGTGG
TGGTAGAAATTGGAGTATTCCCTCTCATTTGTGGTTGGTGGCTGGATATCTGTTCCTTGGAAAT
GTTTGATGCTACTCTGAAAGATCGAGAACTGAGCTTTCAGTCGGCTCCAGGTACTACCATGTTT
CTGCATTGGCTAGTGGGAATGGTATATGTCTTCTACTTTGCCTCCTTCATTCTACTACTGAGAG
AGGTACTTCGACCTGGTGTCCTGTGGTTT

>1 (CU12IR)

TAACCCACCAGCTGAGAACGCAGTGGTGGGGGAAAACCCTGATGCCCAGGATGACCAGGCAGAA
GAGGAGGAGGAGGACAATGAGGAGGAAGATGACGCTGGTGTGGAGGATGCGGCAGATGCTAATA
ACGGAGCCCAGGATGACATGAATTGGAATGCTTTAGAATGGGACCGAGCTGCTGAAGAGCTTAC
ATGGGAAAGAATGCTAGGACTTGATGGATCACTAGTTTTTCTGGAACATGTCTTCTGGGTGGTA
TCTTTAAATACACTGTTCATTCTTGTTTTTGCATTTTGCCCTTACCATATTGGTCATTTCTCCC
TTGTTGGTTTGGGATTTGAAGAACACGTCCAAGCATCTCATTTTGAAGGCCTAATCACAACCAT
AGTTGGGTATATACTTTTAGCAATAACACTGATAATTTGTCATgtatcctttaatgaaccaact
tgggtgtacactaatattattcataccaaagaactcttcattaatggcgaaacttgtctatcct
tgtcctcttagtaaaagttgtaggtatttgcagtaagggtcagtgtggacaaaatgtgaaataa
attaattctgtcaaaaaatactaaatgttggctaagtagttacctgtttcattgtacctttaaa
tactcagtttttccttgacctgatgctgtagGGCTTGGCAACTCTTGTGAAATTTCATAGATCT
CGTCGCTTACTGGGAGTCTGCTATATTGTTGTTAAGGTCTCTTTGTTAGTGGTGGTAGAAATTG
GAGTATTCCCTCTCATTTGTGGTTGGTGGCTGGATATCTGTTCCTTGGAAATGTTTGATGCTAC
TCTGAAAGATCGAGAACTGAGCTTTCAGTCGGCTCCAGGTACTACCATGTTTCTGCATTGGCTA
GTGGGAATGGTATATGTCTTCTACTTTGCCTCCTTCATTCTACTACTGAGAGAGGTACTTCGAC
CTGGTGTCCTGTGGTTT

RABL2B

>Normally spliced transcript (NM_001130923.3). Nucleotide sequence extends from exon 3 to exon 7.

GATCATCTGCCTGGGAGACAGCGCAGTGGGCAAATCCAAACTCATGGAGAGATTTCTCATGGAT
GGCTTTCAGCCACAGCAGCTGTCCACGTACGCCCTGACCCTGTACAAGCACACAGCCACGGTAG
ATGGAAGGACCATCCTTGTGGACTTTTGGGACACGGCAGGCCAGGAGCGGTTCCAGAGCATGCA
TGCCTCCTACTACCACAAGGCCCACGCCTGCATCATGGTGTTTGA

>1 (CU12IR)

GATCATCTGCCTGGGAGACAGCGCAGTGGGCAAATCCAAACTCATGGAGAGATTTCTCATGGAT
GGCTTgtatccttcaaggtttgaagtacccctgttcctgtgggtcttcccacgctcatgtatc
agtgtcccactgcccacacctttgtaccagcagcccaggaaggaggattgggtaagtgaacgca
caggttttgttccgcacttgtttcttgccgattgacccacatggcattgcacaatcagtgctgt
atattttggaccttactcatatttggattgttttatattaagagggcagtagaatggcaaacag
caaatggtttggatctaatgcctggattcaaaccccacctcctcctcctcagtagcttctgata
ctggccaagtcatttaacctctctaacctcaaagcttgggccccctactattgaaaaaaatctc
tgatatgaatgaactttgtaaaataggagctgccagccccacctttctcccatgaattgttata
ttacataggtttggagtgagttcctccccgcaactgggagatttttaaacagcttcccaagtgg
atccactgagaaccacaggtgagaggagcccctggagcctgctgctgcctgtcctaggctcggc
accccaaggctcagttgccttaagatctgagtatgtgttactccaccttgtaccaagtgctcct
gcacacccagcccacctgcgtcatttggctctgctttttacactgtccggcttcttgcagcctc
ggaagctgttaatccccagagtttaacccttttctcagggtttcacacatagacagactccaa
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ccacatttgcattttccaactgcatgatgccttttatgacacttgtctgtaaaatcctgttctg
tgttttacctcagacttcgatggccaggacacactcccatctcctcccacccctcacccttcca
tggagtttgtaccagtgatatgtgaagctgatcctctgtctgcaacattcatttgatttaaaat
ctcaataactccaaagagaagaggagggagaggatccatttcatcaaaccaaacacccctgctc
cccactccttccttacctccccttttattgattttgctccttaacctgagtgcagTCAGCCACA
GCAGCTGTCCACGTACGCCCTGACCCTGTACAAGCACACAGCCACGGTAGATGGAAGGACCATC
CTTGTGGACTTTTGGGACACGGCAGGCCAGGAGCGGTTCCAGAGCATGCATGCCTCCTACTACC
ACAAGGCCCACGCCTGCATCATGGTGTTTGA

>2 (A5SS & A3SS)

GATCATCTGCCTGGGAGACAGCGCAGTGGGCAAATCCAAACTCATGGAGAGATTTCTCATGGAT
GGCTTgtatccttcaaggtttgaagtacccctgttcctgtgggtcttcccacgctcatgtatc
agtgtcccactgcccacacctttgtaccagcagcccaggaaggaggattggacttcgatggcca
ggacacactcccatctcctcccacccctcacccttccatggagtttgtaccagtgatatgtgaa
gctgatcctctgtctgcaacattcatttgatttaaaatctcaataactccaaagagaagaggag
ggagaggatccatttcatcaaaccaaacacccctgctccccactccttccttacctccccttttt
attgattttgctccttaacctgagtgcagTCAGCCACAGCAGCTGTCCACGTACGCCCTGACCC
TGTACAAGCACACAGCCACGGTAGATGGAAGGACCATCCTTGTGGACTTTTGGGACACGGCAGG
CCAGGAGCGGTTCCAGAGCATGCATGCCTCCTACTACCACAAGGCCCACGCCTGCATCATGGTG
TTTGA

SBNO1

>Normally spliced transcript (NM_001167856.3). Nucleotide sequence extends from exon 7 to exon 12.

TATGCAGCCCAGCAACATGAAACTTTCCTACCTAATGGAGATCGTGCTGGCTTCTTAATAGGTG
ATGGTGCCGGTGTAGGAAAAGGAAGGACGATAGCAGGAATCATCTATGAAAATTATTTGTTGAG
TAGAAAACGAGCATTGTGGTTTAGTGTTTCAAATGACTTAAAGTATGATGCTGAAAGAGATTTA
AGGGATATTGGAGCAAAAAACATTTTGGTTCATTCGTTAAATAAGTTTAAATACGGAAAAATTT
CTTCCAAACATAATGGGAGTGTGAAAAAGGGTGTTATTTTTGCTACTTACTCTTCACTTATTGG
TGAAAGCCAGTCTGGCGGCAAGTATAAAACTAGGTTAAAACAACTTCTGCATTGGTGCGGTGAT
GACTTCGATGGAGTGATAGTGTTTGATGAGTGTCATAAAGCCAAAAACTTATGTCCTGTTGGTT
CTTCAAAGCCAACCAAGACAGGCTTAGCAGTTTTAGAGCTTCAGAACAAATTGCCAAAAGCCAG
AGTTGTTTATGCTAGTGCAACTGGTGCTTCTGAACCACGCAACATGGCCTATATGAACCGTCTT
GGCATATGGGGTGAGGGTA

>1 (CU12IR)

TATGCAGCCCAGCAACATGAAACTTTCCTACCTAATGGAGATCGTGCTGGCTTCTTAATAGGTG
ATGGTGCCGGTGTAGGAAAAGGAAGGACGATAGCAGGAATCATCTATGAAAATTATTTGTTGAG
TAGAAAACGAGCATTGTGGTTTAGTGTTTCAAATGACTTAAAGTATGATGCTGAAAGAGATTTA
AGGGATATTGGAGCAAAAAACATTTTGGTTCATTCGTTAAATAAGTTTAAATACGGAAAAATTT
CTTCCAAACATAATGGGAGTGTGAAAAAGGGTGTTATTTTTGCTACTTACTCTTCACTTATTGG
TGAAAGCCAGTCTGGCGGCAAGTATAAAACTAGGTTAAAACAACTTCTGCATTGGTGCGGTGAT
GACTTCGATGGAGTGgtatcctttatggcaaatatttttctatttttaacgacaagaaaatta
taaaagggtagacctttgggaaacaggaatcttgagattaaacgagttgctggagtgaaaactt
taatgattacctagagtccaaagggattggaaagttgattcggtttttacctgtcagataggа
gaccaatgtttaatctcctggtttagaaaaatgttaatagatatgaggaatactgaatactgta
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gaaaactgttagaacttagtggtcccaaacttgttcacagtatatcttttaactgtcctgcatg
aggaacatcacttttgaaatgctgattaagttggttaccaatcatgcttctacttttacaagt
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tggtttaaattccaaattcagtgaatgtctttgaggctagagagggaggaggatgtaactgggg
aaaggcccataggacttcctttgtatttgcagtcatgaaataggcaatcagaaggcctcaggga
aataaaagttaactcttcttgaaagactggagattcttacttaatactaatgtgatacattcct
taactcattccatttagATAGTGTTTGATGAGTGTCATAAAGCCAAAAACTTATGTCCTGTTGG
TTCTTCAAAGCCAACCAAGACAGGCTTAGCAGTTTTAGAGCTTCAGAACAAATTGCCAAAAGCC
AGAGTTGTTTATGCTAGTGCAACTGGTGCTTCTGAACCACGCAACATGGCCTATATGAACCGTC
TTGGCATATGGGGTGAGGGTA

SLC12A4

>Normally spliced transcript (NM_005072.5). Nucleotide sequence extends from exon 9 to exon 12

AAGGAGAGCCTGCCTCTGTACGTGGTCGCTGACATCGCCACATCCTTCACCGTGCTGGTCGGCA
TCTTCTTCCCTTCTGTAACAGGCATCATGGCTGGCTCAAACCGCTCTGGGGACCTTCGTGACGC
CCAGAAGTCTATCCCTGTGGGGACCATTCTGGCCATCATTACAACTTCCCTCGTGTACTTCAGC

AGTGTGGTTCTCTTTGGTGCCTGCATTGAGGGTGTGGTTCTCCGGGACAAGTATGGCGATGGTG
TCAGCAGGAACT

>1 (CU12IR)

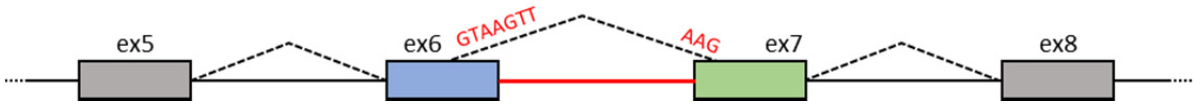
AAGGAGAGCCTGCCTCTGTACGTGGTCGCTGACATCGCCACATCCTTCACCGTGCTGGTCGGCA
TCTTCTTCCCTTCTGTAACAGGCATCATGGCTGGCTCAAACCGCTCTGGGGACCTTCGTGACGC
CCAGAAGTCTATCCCTGTGGGGACCATTCTGGCCATCATTACAACTTCCCTCGTGTgtatcctt
tcccaggcctggggcaggtggggcagaggacgggaagcgcttggcctgtgtacgttgccaggtc
acatgcacagacaccccacacactcatatatagctccccttctcatgcacacatgcacatacac
accacgagactcccagctgtaggtgtcctggtagtctggctggggtgggggcacgtggtactat
tcagggacagtggggtggcagaggccgagttttccttgacgccgccgcagACTTCAGCAGTGTG
GTTCTCTTTGGTGCCTGCATTGAGGGTGTGGTTCTCCGGGACAAGTATGGCGATGGTGTCAGCA
GGAACT

ZC3H8

>Normally spliced transcript (NM_032494.3). Nucleotide sequence extends from exon 4 to exon 9.

GCTGGTCACAAGAATGGCAAACAGAAGAAAATGAAGCGAAAATGGCCTGGCCCTGGAAACAAAG
GATCAAATGCTTTGCTGAGGAACAGCGGCTCACAGGAAGAGGATGGTAAACCTAAAGAGAAGCA
GCAGCATTTGAGTCAGGCATTCATCAACCAACATACAGTGGAACGCAAGGGAAAACAAATTTGT
AAATATTTTCTTGAAAGGAAATGTATTAAGGGAGACCAGTGTAAATTTGATCATGATGCAGAGA
TAGAAAAGAAAAAGGAAATGTGTAAGTTTTATGTACAAGGATATTGTACCAGAGGTGAAAACTG
TCTGTATTTGCATAATGAATATCCTTGTAAGTTTTACCATACAGGAACAAAATGTTATCAGGGA
GAATACTGCAAGTTTTCTCATGCTCCACTGACTCCTGAAACACAAGAATTGTTGGCTAAAGTTT
TGGATACTGAAAAGAAGTCATGTAAATAAAATAGACATAAAAAGGTAGCAATGTACAGATAAAG
AGTACTTTAACGCCCATGCGTGTTCAAGACTGT

>1 (A5SS & A3SS)



GCTGGTCACAAGAATGGCAAACAGAAGAAAATGAAGCGAAAATGGCCTGGCCCTGGAAACAAAG
GATCAAATGCTTTGCTGAGGAACAGCGGCTCACAGGAAGAGGATGGTAAACCTAAAGAGAAGCA
GCAGCATTTGAGTCAGGCATTCATCAACCAACATACAGTGGAACGCAAGGGAAAACAAATTTGT
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AGTTTTCTCATGCTCCACTGACTCCTGAAACACAAGAATTGTTGGCTAAAGTTTTGGATACTGA
AAAGAAGTCATGTAAATAAAATAGACATAAAAAGGTAGCAATGTACAGATAAAGAGTACTTTAA
CGCCCATGCGTGTTCAAGACTGT