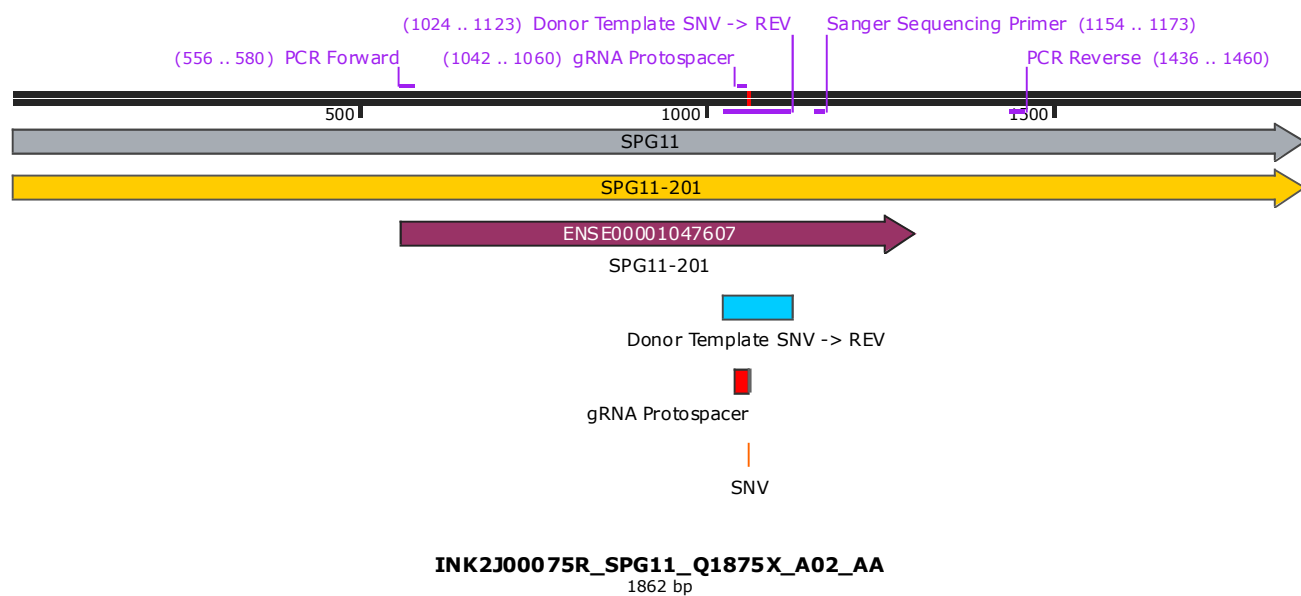


SPG11 vesicle trafficking associated, spatacsin [Source:HGNC Symbol;Acc:HGNC:11226]



5' TTGTAGGATATGCTACAAC TATTTTCTTTGAATAGAATTA AATATAAGGGGGCTTCCACTTAAACCCCATTC CATGAGATCAAT 85
 3' AACATCCTATACGATGTTGATAAAAAGGAAACTTATCTTAATTTATATTCCCCGAAGGTGAATTTGGGGTAAGGTACTCTAGTTA
 SPG11
 SPG11-201

CAAGAATTGTTTTTCATACCATAACTTTCCAGATAAAATATCTAGGGTCATCTGTAGCCAAATCTCTGTTCTCTGATTTGATTTG 170
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 SPG11
 SPG11-201

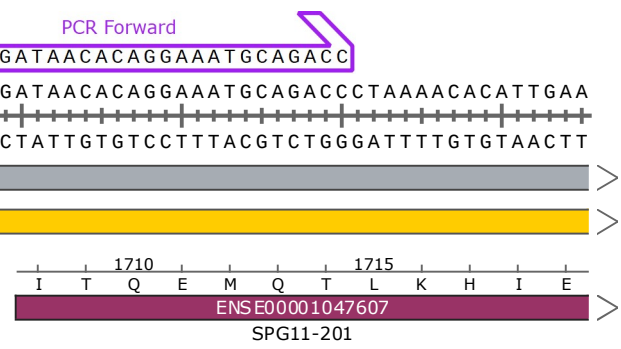
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 SPG11
 SPG11-201

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 SPG11
 SPG11-201

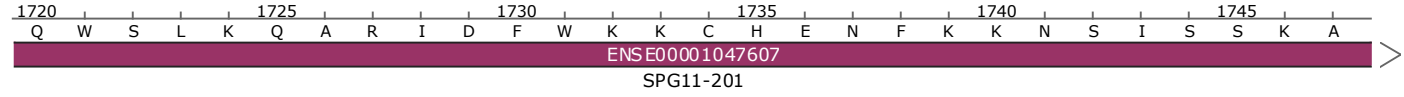
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 SPG11
 SPG11-201

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 SPG11
 SPG11-201

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 SPG11
 SPG11-201



CAGTGGTCACTAAAACAAGCAAGAATTGACTTCTGGAAAAAATGCCATGAGAATTTAAGAAAAATTCAATTTCAAGCAAAGCAG 680
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 SPG11
 SPG11-201



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SPG11
 SPG11-201

1750 1755 1760 1765 1770 1775
 A S S F F S T Q A H V A C E H P T G W S S M E E R H L L L

ENSE00001047607
 SPG11-201

CACCTTGGCAGGGCACTGGCTTGCCAGGAGGACGTGGTGCCTTGGATAAGCTGGAGGAGCTGGAGAAGCAGATCTGGCTGTGC
 GTGGAACCGTCCCGTGACCGAACGGGTCTCTGCACCACGGGAACCTATTGCACCTCCTCGACCTCTTCGTCTAGACCGACAG

SPG11
 SPG11-201

1780 1785 1790 1795 1800
 T L A G H W L A Q E D V V P L D K L E E L E K Q I W L C

ENSE00001047607
 SPG11-201

CGCATCACCCAGCACACTCTTGAAGAAATCAGGAGGAAACAGAGCCCAGATTTTCTCGACAGATCTCAACTAGTGGTGAACTTT
 GCGTAGTGGGTCTGTGTGAGAACCTTCTTTAGTCCTCTTGTCTCGGGTCTAAAAGAGCTGTCTAGAGTTGATCACCACCTTGAAA

SPG11
 SPG11-201

1805 1810 1815 1820 1825 1830
 R I T Q H T L G R N Q E E T E P R F S R Q I S T S G E L

ENSE00001047607
 SPG11-201

CCTTTGATAGTTTAGCCAGTGAGTTTTCTTCTCCAAGTTGGCTGCTCTGAACACATCAAATACTTAGAACTTAACAGCCTTCC
 GGAAACTATCAAATCGGTCACTCAAAGGAAGAGGTTCAACCGACGAGACTTGTGTAGTTTTATGAATCTTGAATTGTCGGAAGG

SPG11
 SPG11-201

1835 1840 1845 1850 1855 1860
 S F D S L A S E F S F S K L A A L N T S K Y L E L N S L P

ENSE00001047607
 SPG11-201

gRNA Protospacer
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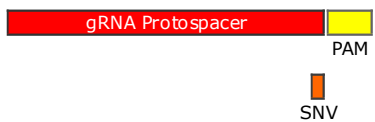
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SPG11
 SPG11-201

1865 1870 1875 1880 1885
 S K E T C E N R L D W K E Q E S L N F L I G R L L D D G

ENSE00001047607
 SPG11-201

Donor Template SNV -> REV



GTTTCTCTGTACGCTCTTATCTAACCTAACCTTTCTCGTCTCAGTGATTTGAAAACTAACC CGGGATGACCTACTACCG
 Donor Template SNV -> REV

TGTGTGCATGAAGCAAGTAGAGTATGCCGGTATTTTCATTTTTATAATCCAGATGTCGCCTTGGTATTGCACTGCAGAGCACTGG
 ACACACGTA CTTCGTTTCATCTCATACGGCCATAAAAAGTAAAAATATTAGGTCTACAGCGGAACCATAACGTGACGTCTCGTGACC

SPG11
 SPG11-201

1890 1895 1900 1905 1910 1915
 C V H E A S R V C R Y F H F Y N P D V A L V L H C R A L

ENSE00001047607
 SPG11-201

Donor Template SNV -> REV

ACACACGTA CTTCGTTCA
 Donor Template SNV -> REV

GGTCTACAGCGGAACCATAA
 Sanger Sequencing Primer

CCTCAGGGGAAGCTAGTATGGAGGATCTGCACCCAGAGATCCATGCTCTCCTACAAAGTGCTGAGCTGCTTGAGGAAGAAGCACC
 GGAGTCCCCTTCGATCATACCTCCTAGACGTGGGTCTCTAGGTACGAGAGGATGTTTCACGACTCGACGAACCTCTTCTTCGTGG

SPG11
 SPG11-201

1920 1925 1930 1935 1940 1945
 A S G E A S M E D L H P E I H A L L Q S A E L L E E E A P

ENSE00001047607
 SPG11-201

CGACATCCCCTAAGGAGAGTCCACAGCAGTAAGTGAAGGAGATCAGATGGCCCAGAGTCTTAAATGGCACTGTTAGCACTGTCT
 GCTGTAAGGGGATTCTCTCAGGTGTCGTCATTCCTCTAGTCTACCGGTCTCAGAATTTACCGTGACAATCGTGACAGA

SPG11
 SPG11-201

1950 1955
 D I P L R R V H S S K *

ENSE00001047607
 SPG11-201

CCAAGAAGGAAGGGACCCTTGTGGCAGAATAGTTCTACCAAGTTAAGGGGACAACAACCTTTTTCCAGGTAATCTGTAGACTAGC
 GGTTCTTCCCTGGAACACCGTCTTATCAAGATGGTTCAATCCCTGTTGTTGAAAAAAGGTCCATTAGACATCTGATCG

SPG11
 SPG11-201

CATCTGATCG
 PCR Reverse

CAATTAAGTGGCAATGAAATAGAAAGATCTATGTCTTTTAACTAGCTATGTTTCAAAGTTAAAGGGAATCTATCTGATTTGTA
 GTTAATTGATCCGTTACTTTATCTTTCTAGATACAGAAAATTTGATCGATACAAAGTTTCAATTTCCCTTAGATAGACTAAACAT

SPG11
 SPG11-201

GTTAATTGATCCGTT
 PCR Reverse

GCTAGTTCACTGAAATTTATTCTACATATTCTATACATCTGTAAATTTAATCATTTCCAGTCTTACATTGATAGTATATAGAAAT
 CGATCAAGTGACTTTAAATAAGATGTATAAGATATGTAGACATTTAAATTAGTAAAGGTCAGAATGTAACATCATATATCTTTA

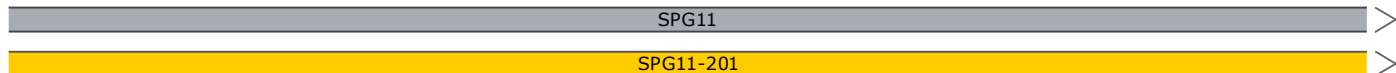
SPG11
 SPG11-201

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SPG11
 SPG11-201

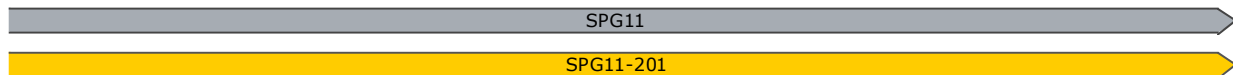
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1785



GCTAATTTTTGTATTAGTAGAGACCGGGTTTCACCATGTTGGCCAGGCTGGTCTCGAACTCCTGACCTCAGGCGATT
 CGATTAAAAACATAATCATCTCTGGCCCAAAGTGGTACAACCGGTCCGACCAGAGCTTGAGGACTGGAGTCCGCTAA

3 '
 1862
 5 '



Feature	Location	Size	±	Type
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✓ SPG11-201	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000261866				
SPG11-202	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000427534				
SPG11-203	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000535302				
SPG11-209	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000558319				
SPG11-216	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000559511				
SPG11-220	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000561391 Retained intron				
SPG11-221	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000682065				
SPG11-223	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000682460 Nonsense mediated decay				
SPG11-224	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000682495 Nonsense mediated decay				
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/note = primary transcript ENST00000682669				
SPG11-231	1 .. 1862	1862 bp	☐ →	prim_transcript
/note = primary transcript ENST00000683186 Nonsense mediated decay				
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/note = primary transcript ENST00000684676 Nonsense mediated decay				
SPG11-211	1 .. 859	859 bp	☐ →	prim_transcript
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✓ SPG11-201	560 .. 1304	745 bp	☐ →	CDS
/note = coding sequence ENSP00000261866				
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Feature	Location	Size	Start	End	Type
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/note	= coding sequence ENSP00000396110				
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SPG11-203	560 .. 1304	745 bp	■	→	CDS
/note	= coding sequence ENSP00000445278				
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SPG11-209	560 .. 1304	745 bp	■	→	CDS
/note	= coding sequence ENSP00000453599				
/translation	= ITQEMQTLKHIEQWSLKQARIDFWKKCHENFKKNSISSKAASSFFSTQAHVACEHPTGWSSMEERHLLLTLAGHWLAQEDVVPLDKLEELEKQIWLCRITQHTLGRNQEETEP RFSRQISTSGELSFDSLASEFVSFKLAALNTSKYLELNSLPSKETCENRLDWKEQESLNFLIGRLDDGCVHEASRVCRYFHFYNPDPVALVLHCRALASGEASMEDLHPEIHA LLQSAELLEEEAPDIPLRRVHS 248 amino acids = 28.5 kDa				
SPG11-216	560 .. 1304	745 bp	■	→	CDS
/note	= coding sequence ENSP00000453246				
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SPG11-226	560 .. 1304	745 bp	■	→	CDS
/note	= coding sequence ENSP00000507782				
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SPG11-239	560 .. 1304	745 bp	■	→	CDS
/note	= coding sequence ENSP00000508295				
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SPG11-221	704 .. 1304	601 bp	■	→	CDS
/note	= coding sequence ENSP00000507025				
/translation	= AHVACEHPTGWSSMEERHLLLTLAGHWLAQEDVVPLDKLEELEKQIWLCRITQHTLGRNQEETEP RFSRQISTSGELSFDSLASEFVSFKLAALNTSKYLELNSLPSKETCENRLDWKEQESLNFLIGRLDDGCVHEASRVCRYFHFYNPDPVALVLHCRALASGEASMEDLHPEIHA LLQSAELLEEEAPDIPLRRVHS 200 amino acids = 22.8 kDa				
✓ Donor Template SNV -> REV	1024 .. 1123	100 bp	■		misc_feature
✓ gRNA Protospacer	1042 .. 1061	20 bp	■		misc_feature
✓ SNV	1061 .. 1061	1 bp	■		misc_feature
/note	= SNV = T REV = C				
✓ PAM	1062 .. 1064	3 bp	■		misc_feature
EIF3J	1863 ..19,555	17,693 bp	■	←	gene
/note	= gene ENSG00000104131 Protein coding				
EIF3J-202	1863 ..19,555	17,693 bp	■	←	prim_transcript
/note	= primary transcript ENST00000424492 Protein coding				
EIF3J-201	1863 ..19,533	17,671 bp	■	←	prim_transcript
/note	= primary transcript ENST00000261868 Protein coding				
EIF3J-203	1863 ..19,497	17,635 bp	■	←	prim_transcript
/note	= primary transcript ENST00000535391 Protein coding				

Feature	Location	Size	±	Type
EIF3J-205	1863 .. 5242	3380 bp	☐ ←	prim_transcript
/note	= primary transcript ENST00000558053 Retained intron			
EIF3J-204	1863 .. 2056	194 bp	☐ ←	prim_transcript
/note	= primary transcript ENST00000536248 Protein coding			

Primer	Length	Binding Sites	Tm	Date Added
✓ PCR Forward	25-mer	556 .. 580	59°C	Mar 12, 2024
/sequence = ACAGATAACACAGGAAATGCAGACC 44% GC / 7686.1 Da				
✓ Donor Template SNV -> REV	100-mer	1024 .. 1123	76°C	Mar 12, 2024
/sequence = ACTTGCTTCATGCACACAGCCATCATCCAGTAGGCGCCCAATCAAAAAGTTTAGTGACTCCTGCTCTTTCCAATCCAATCTATTCTCGCATGTCTCTTTG 45% GC / 30,458.8 Da				
✓ gRNA Protospacer	20-mer	1042 .. 1060	49°C	Mar 12, 2024
/sequence = TAGATTGGATTGGAAAGAGT 35% GC / 6260.2 Da				
✓ Sanger Sequencing Primer	20-mer	1154 .. 1173	57°C	Mar 12, 2024
/sequence = AATACCAAGGCGACATCTGG 50% GC / 6135.1 Da				
✓ PCR Reverse	25-mer	1436 .. 1460	56°C	Mar 12, 2024
/sequence = TTGCCTAGTTAATTGGCTAGTCTAC 40% GC / 7638.0 Da				