

INK2J00034_CACNA1A_R583Q_C05_BB
20,614 bp

CCTGGGCAACAAAGCAAGATCCTGTCTCAAAAAAAAAAAAAAAAGATGTATTTTAGAAGGTAAATTCAATCTGTCCAAAACTGAG HبץبН ب GGACCCGTTGTTTCGTTCTAGGACAGAGTTTTTTTTTTTTTTTTCTACATAAAATCTTCCATTTAAGTTAGACAGGTTTTGACTC

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CTCTGACCTTCCCCTAAACCTGTGCCCATTCAGTGGATGAGAGCTCCATCCCTTAAGGGGTTCACCAATTCATCCATTCCTTTGT
 GAGACTGGAAGGGGATTTGGACACGGGTAAGTCACCTACTCTCGAGGTAGGGAATTCCCCAAGTGGTTAAGTAGGTAAGGAAACA

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ATGTACATCATTCATTCACCTTGGCTCATCCCTCTCTCTTACATCCACACCGTTCCATCAGCAAATGTTGAATCTGTCTTAAATG

TACATGTAGTAAGTAAGTGGAACCGAGTAGGGAGAGAGAATGTAGGTGTGGCAAGGTAGTCGTTTACAACTTAGACAGAATTTAC

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ATTCATCCCAAATCCTCCCCGCTTAACTACCACCCAACTCCAGCCCCCATCCATCATCATCATCACTTGCCTGGATGGGTTCAGT
 TAAGTAGGGTTTAGGAGGGGCGAATTGATGGTGGGTTGAGGTCGGGGGTAGGTAGTAGTAGTAGTGAACGGACCTACCCAAGTCA

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CACCTCCAGCCTGGTCTCCCAGCTCCCGTCCTCACCTCTCACTGTCTACTCTCCCACTCGGCAGCCAGAGGGTGCCTGTGAACAC
 GTGGAGGTCGGACCAGAGGGTCGAGGGCAGGAGTGGAGAGTGACAGATGAGAGGGTGAGCCGTCGGTCTCCCACGGACACTTGTG

| $\square$ |
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TCCACCAGGCCATGCATGATCTGCCTGTCACCTCCCTGCCTTCACCACCTTCCTCTTTTCCCCTCAACCACTCCACTCCAGCCAC

AGGTGGTCCGGTACGTACTAGACGGACAGTGGAGGGACGGAAGTGGTGGAAGGAGAAAAGGGGAGTTGGTGAGGTGAGGTCGGTG

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ACTGACTTCCTTGTGCTCTTCCCCAAAAATGTCGGGCAGACACATTCATGCTTCAGGACCTTAAATTTGCTGTTTCCTCTACCTA
 TGACTGAAGGAACACGAGAAGGGGTTTTTACAGCCCGTCTGTGTAAGTACGAAGTCCTGGAATTTAAACGACAAAGGAGATGGAT

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AGATACTAAAGTGACAAGTCAACACACTCACCTTGACCATGCAATTTAATGTTGCAGCCTACCCTGTGGACTCTCCAAGGGCTCC
 TCTATGATTTCACTGTTCAGTTGTGTGAGTGGAACTGGTACGTTAAATTACAACGTCGGATGGGACACCTGAGAGGTTCCCGAGG

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CAGTCCCTCTGTGATGCTTTACTTTTTCTCTTAAAAAAAAAATTGTTATTTAAAAGAACTTGTCTCGCTGTGTTGCCCAGGCTGG
 GTCAGGGAGACACTACGAAATGAAAAAGAGAATTTTTTTTTTAACAATAAATTTTCTTGAACAGAGCGACACAACGGGTCCGACC

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TGTCAAACTCCTGGCCTCATACAGTCCTCCCATTCCAGCTTCCCAAAGTACTGGGATTAGAGGCATGTGCCACTGCACCCATCCC
 ACAGTTTGAGGACCGGAGTATGTCAGGAGGGTAAGGTCGAAGGGTTTCATGACCCTAATCTCCGTACACGGTGACGTGGGTAGGG

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 TTGAAAAAAAAAGGGTATCGTGAAAAGTAAAAGGTAGGGTGACAATTAAATGAATAATGCAGGTGACAGACAGAGGAGGGGAATC

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 ATTTACGTATTCTCGTTTTGATATACATCCGTCTCCTGTGTGGGTCGAATAAGGAGTCACTAGTGAAGATTTCAATTTACAGGTA

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GGAAAACAGTCTCATCCACATCTCTTTCTGGAGGCCTTCCAAGCGTGCTCCATGCAGCTCTGTTGCCTGCCCCTGCATCAGGGAA
 CCTTTTGTCAGAGTAGGTGTAGAGAAAGACCTCCGGAAGGTTCGCACGAGGTACGTCGAGACAACGGACGGGGACGTAGTCCCTT

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TGGAGGCTCTGCTTTATCCTGCCCTGTGGTGTGACTCCCAGAGGCATCAGATGTGGCTGGGAGTGGGAGACATGGAAAATTGGCT
 ACCTCCGAGACGAAATAGGACGGGACACCACACTGAGGGTCTCCGTAGTCTACACCGACCCTCACCCTCTGTACCTTTTAACCGA


CCTGCAACAGAGAACTATCAGCCTTCCCATCAATTGGTTACTTCTAATTCTGTTATTTTTCAGGGGCACTGTCTTCTCATAAGCT
 GGACGTTGTCTCTTGATAGTCGGAAGGGTAGTTAACCAATGAAGATTAAGACAATAAAAAGTCCCCGTGACAGAAGAGTATTCGA

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CCATCTATGCAAAACTAAGCCCATGGGTCATGATGGTTCCCTCAGGCCAGAGGCTTGCTGGAGAGACTAATGGATCCCCTGGCTA
 GGTAGATACGTTTTGATTCGGGTACCCAGTACTACCAAGGGAGTCCGGTCTCCGAACGACCTCTCTGATTACCTAGGGGACCGAT

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AAATCTGTGCTTGGGCTGCACATTGGTTAATTTCTTCTGAAGGAACAGCCTGAGCCTGACATTCTCCATCTTTTCCCTGGCAGGT بमبН TTTAGACACGAACCCGACGTGTAACCAATTAAAGAAGACTTCCTTGTCGGACTCGGACTGTAAGAGGTAGAAAAGGGACCGTCCA


TCTCCCTTCGCCCGAGCCAGCATTAAAAGTGCCAAGCTGGAGAACTCGACCTTTTTTCACAAAAAGGAGAGGAGGATGCGTTTCT ب中 ب ب ب ب AGAGGGAAGCGGGCTCGGTCGTAATTTTCACGGTTCGACCTCTTGAGCTGGAAAAAAGTGTTTTTCCTCTCCTCCTACGCAAAGA


ACATCCGCCGCATGGTCAAAACTCAGGCCTTCTACTGGACTGTACTCAGTTTGGTAGCTCTCAACACGCTGTGTGTTGCTATTGT
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TCACTACAACCAGCCCGAGTGGCTCTCCGACTTCCTTTGTGAGTATCACCCAGCCCCACCCCTGCCAACTCCCTGATCCCTCCCT
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CACACCCTTTTTCCACTTCTCTTTCTCTGGTAGTATGTGTATCTTCTTTGGTCCTCATTGAATCTGCCCTTTCCTTTAGCCATTT
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CTATAACTGTCACTGGGGCCAATGTTACTGTTGCTATGACAATGGAACCCATCTCCCTTAGACCTGAGAGCTGGAAGCTGGAATT ННН GATATTGACAGTGACCCCGGTTACAATGACAACGATACTGTTACCTTGGGTAGAGGGAATCTGGACTCTCGACCTTCGACCTTAA


CAGACCAACAAATGCTCCTGTGATTCCTTTCTAAGAGAGAGGGACAGAGGGGTGCTGGTGAAGGGGATGTTGGAAGAGAGACAGA
 GTCTGGTTGTTTACGAGGACACTAAGGAAAGATTCTCTCTCCCTGTCTCCCCACGACCACTTCCCCTACAACCTTCTCTCTGTCT


GAAAGACGGAGCTCATAAGATAGACAGATAGAAACAGAAACATACATGTATTAATAATTTTTATGTACATCTCTGGAAATGTTCA HبНبН CTTTCTGCCTCGAGTATTCTATCTGTCTATCTTTGTCTTTGTATGTACATAATTATTAAAAATACATGTAGAGACCTTTACAAGT

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TTCTCTCTTCTTTCAAACCTTCTGCTTCTTTCCTGTTAGGTTGGTGCAAAATTAATTGCGTTTTTTGCCTTTTTTTTTTTTTTT
 AAGAGAGAAGAAAGTTTGGAAGACGAAGAAAGGACAATCCAACCACGTTTTAATTAACGCAAAAAACGGAAAAAAAAAAAAAAAA


TTAACCACAGTTACTTTTGCACCAACCTAATACTTCCTCCCCTGCCCTTTTTGGCTTCCTTATTCATTCATAGAACATCCCCTCC
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AGTATCTGCGAGAGCGTTTTGCTCCCTCAAGGTACAAGGCCCACTAAGGCTTTGCCCTCTGGGCCTATTCCCAGATTCTATGTGA بнНبץبץبץ TCATAGACGCTCTCGCAAAACGAGGGAGTTCCATGTTCCGGGTGATTCCGAAACGGGAGACCCGGATAAGGGTCTAAGATACACT



AAAAAGTAAAGAGCTCGAGGAATCAACAAGAGCAGCGACTGGGGCCAGGCATGGTGGCTCACACCTGTAATCCCAGCATTTTGGG

TTTTTCATTTCTCGAGCTCCTTAGTTGTTCTCGTCGCTGACCCCGGTCCGTACCACCGAGTGTGGACATTAGGGTCGTAAAACCC


AGGCTGAGGTGGGTGGATCACTTGAGGCCAGGAGTTCAAGACCAGCCTGGCCAATATGGTGAAACCCTGTCTTTACAAAAAATAC

TCCGACTCCACCCACCTAGTGAACTCCGGTCCTCAAGTTCTGGTCGGACCGGTTATACCACTTTGGGACAGAAATGTTTTTTATG


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taAAATAAAATAAAATAAAGAGTAGTGATtGgGCAGTGAGGGGGGCAGGTGGATGCCCTGGCTTTGGCTCACAGGCcCCAAGTAA
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GTCACGTTGGCTGACCCCCCAGGACACCCCTCAGGAACCAGTTCTCCTTCCCAGGGCCCTGACCTAGTTTCAAACTTAGTAATTG بН $ب+\boldsymbol{H}$ +
CAGTGCAACCGACTGGGGGGTCCTGTGGGGAGTCCTTGGTCAAGAGGAAGGGTCCCGGGACTGGATCAAAGTTTGAATCATTAAC

tTtTTAGTCCCTCTGGAGTCTCTTATAAATGAGGACTCTACTTCGTGTTTTAACTTCCTCTAATACTCTATTTTTAATCTCCTAT

AAAAATCAGGGAGACCTCAGAGAATATTTACTCCTGAGATGAAGCACAAAATTGAAGGAGATTATGAGATAAAAATTAGAGGATA


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CAGTGAGCCCAGATCATGCCACTGCACTCTAGCCTGGGCAACAGAGCTAGACGCCATCTCAAAAAAAAAAAAAAAAAAAAAAGA
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TACACATTAATTTCAGAGATGTCAAAATATAAACAAAAATGTATATCTTGGCATCAGTGAAGTGTAGTTGTTTCTCTGGATCTCA بम+ ATGTGTAATTAAAGTCTCTACAGTTTTATATTTGTTTTTACATATAGAACCGTAGTCACTTCACATCAACAAAGAGACCTAGAGT


GACTCCACATCTATGTGGTAGAAACCGGATTTGATGGTCCTGAAAGTTCTTCCAGATGCAACAATGCTAAGGATAAGTAATTCTT بн+ ب + + CTGAGGTGTAGATACACCATCTTTGGCCTAAACTACCAGGACTTTCAAGAAGGTCTACGTTGTTACGATTCCTATTCATTAAGAA


TCAAGTCTTGTGCATCACCTGCTATCATGTTTCCATGGTAACTGAGGAACAAGATCTCAGAAACTCTTCAGTCCTCCCAGAGTTA

AGTTCAGAACACGTAGTGGACGATAGTACAAAGGTACCATTGACTCCTTGTTCTAGAGTCTTTGAGAAGTCAGGAGGGTCTCAAT


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AAACACCTCTTAAAAGCATGACATAGCAAACACCCTTGGCAAATATCTTAGTTCATTTGTACTGCTATAACAAATTACCCGAGAC
 TTTGTGGAGAATTTTCGTACTGTATCGTTTGTGGGAACCGTTTATAGAATCAAGTAAACATGACGATATTGTTTAATGGGCTCTG


TGGGTAATTTGATAAGAACAGAAATTTATTTTCTCACAGTTCTGGAGGCTGGGAAGCCCAAGATCAAGGCATTGGCAGGTTTCCC بץبץب ACCCATTAAACTATTCTTGTCTTTAAATAAAAGAGTGTCAAGACCTCCGACCCTTCGGGTTCTAGTTCCGTAACCGTCCAAAGGG


TGTCTGGCGAAAGCTACTCTCTGCTTCCAAGATTGCACCTTGAACACTGTATCCTCTGGAAGGGAGGAACACTGGGTCCTTACAT
 ACAGACCGCTTTCGATGAGAGACGAAGGTTCTAACGTGGAACTTGTGACATAGGAGACCTTCCCTCCTTGTGACCCAGGAATGTA


GGCAGAAGGTGGAGGAGCAAGAGGGACAAACTTCCTCTGTCAACCTCTTTTATAAGGGCACCTAATCCCATTCATGAGAGCTCTA HبH CCGTCTTCCACCTCCTCGTTCTCCCTGTTTGAAGGAGACAGTTGGAGAAAATATTCCCGTGGATTAGGGTAAGTACTCTCGAGAT


CCGTAATGACTTAATCACCTCCTGAAGGCCCCACCTCTTAATACTGTTACATTGGCAATTAAGTTTCAACGTGAATTTTGGAGGG



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TTCAGTCCTCTGTCTCTTTCCTTTCTGTATGCTTTCTGCTCCTCAGAAACCCTCCTCATCTCTCCTTTCTATCCATTAAGTACCC世十НبН AAGTCAGGAGACAGAGAAAGGAAAGACATACGAAAGACGAGGAGTCTTTGGGAGGAGTAGAGAGGAAAGATAGGTAATTCATGGG


ACGCCCTTCCTAACTCCTCATCTTCCTACCCTACCAAGAAAGCCCTCTCAGAAAAGGATCTGATGTCAGCCATTTATTTGCTGGA
 TGCGGGAAGGATTGAGGAGTAGAAGGATGGGATGGTTCTTTCGGGAGAGTCTTTTCCTAGACTACAGTCGGTAAATAAACGACCT

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TCAAGACTTTTTAGGTCACTCACAACACAGGATTTCTAGGGGACATAAGACAAGTTTTCTGAGTTAGGAGAAAAGCCATACCTTA
 AGTTCTGAAAAATCCAGTGAGTGTTGTGTCCTAAAGATCCCCTGTATTCTGTTCAAAAGACTCAATCCTCTTTTCGGTATGGAAT


GgTGGGTTGCCTGTGTCGCTCCAACTAAGTACTTAACTTCAGGATTACAAATAGGATATCATTATGATTTCTATTTCCTTTTATC H+H H H H
CCACCCAACGGACACAGCGAGGTTGATTCATGAATTGAAGTCCTAATGTTTATCCTATAGTAATACTAAAGATAAAGGAAAATAG


CTTTGGAGCTCAGTCACGTAGAAGTAGATTAAATATAATTGTTAGATCACAGCACCCTGGCATTATGGGGCCGTTATGGTCCATT



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TTAAAATCGGCCTAAGGTTGGTTAGCTTGTTGGTTGGAGGGTAGGGCATAATTGTTGCTTTTTTTTTTTTTTTTTTTTTTAGAC
 AATTTTAGCCGGATTCCAACCAATCGAACAACCAACCTCCCATCCCGTATTAACAACGAAAAAAAAAAAAAAAAAAAAAAATCTG


AAGGTCTTGCTCTGTCACCCAGGCTACAGTAGGGTGGCCCAATCTTGGCTCACTGCAACCTCCACCTCCCAGGTTTAAGTGATTC
 TTCCAGAACGAGACAGTGGGTCCGATGTCATCCCACCGGGTTAGAACCGAGTGACGTTGGAGGTGGAGGGTCCAAATTCACTAAG


TCATGCCTCAGCCTCCCAAGTAGCTGGGTTTACAGGCATGTGTCACCACACTGGCTAATTTTTGTATTTTTAGTAGAGGCGGGGT
 AGTACGGAGTCGGAGGGTTCATCGACCCAAATGTCCGTACACAGTGGTGTGACCGATTAAAAACATAAAAATCATCTCCGCCCCA


TTGCCATGTTAGCCAGGCTGGTCTCAAACTCCTGACCTCAGTTGATCTGACCGCCTAGGCCTCCCAAAGTGCTGGGATTACAGAC

AACGGTACAATCGGTCCGACCAGAGTTTGAGGACTGGAGTCAACTAGACTGGCGGATCCGGAGGGTTTCACGACCCTAATGTCTG


GTGAGCCACCATGCCCAGCCAGCTCTTCCTTTTTAACAGAGGGGAAACTGAGGCCCATGGGAAGGACACCTTGGACAGGGCGTGG

CACTCGGTGGTACGGGTCGGTCGAGAAGGAAAAATTGTCTCCCCTTTGACTCCGGGTACCCTTCCTGTGGAACCTGTCCCGCACC


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CTGTAGTTCTAGCATGTTGGAGGCCAAATCAGGGAAACTGTTTGAGGCCAGGAGTTTGAAACCAGCCTAACAGCATAGCAAGACC
 GACATCAAGATCGTACAACCTCCGGTTTAGTCCCTTTGACAAACTCCGGTCCTCAAACTTTGGTCGGATTGTCGTATCGTTCTGG


TCATCTCTACAAAAAATAAAAAGTTTAAAAATGATAATAAAAGGAAAGTCAGAGCCACCTGGAACCCCTACCCTCAGCAAGCCTA + $+\boldsymbol{+} \boldsymbol{+}$ AGTAGAGATGTTTTTTATTTTTCAAATTTTTACTATTATTTTCCTTTCAGTCTCGGTGGACCTTGGGGATGGGAGTCGTTCGGAT


ACCTCCTCTCTGTTTCCTCCTTCTCCCTTCTAGACTATGCAGAATTCATTTTCTTAGGACTCTTTATGTCCGAAATGTTTATAAA
 TGGAGGAGAGACAAAGGAGGAAGAGGGAAGATCTGATACGTCTTAAGTAAAAGAATCCTGAGAAATACAGGCTTTACAAATATTT
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AATGTACGGGCTTGGGACGCGGCCTTACTTCCACTCTTCCTTCAACTGCTTTGACTGTGGGGTAAGTGCTCTTGTTTCTAAGAGT HبН H TTACATGCCCGAACCCTGCGCCGGAATGAAGGTGAGAAGGAAGTTGACGAAACTGACACCCCATTCACGAGAACAAAGATTCTCA


TCATTTCTCCAGCTCTTGCCTGGAATGACAGATACCTGGACACATTAAAGGGAGAAAGGTAAAGTCACCCCTGAATATGAGAGAC
 AGTAAAGAGGTCGAGAACGGACCTTACTGTCTATGGACCTGTGTAATTTCCCTCTTTCCATTTCAGTGGGGACTTATACTCTCTG



GGAGGAAAACGTATAGACCCCCTTTGGAAAGCTAAGTGGGGGACATAAGACAAGTTTTCCAAGTTGGGAGAAAAGCCATGCCTTA
 CCTCCTTTTGCATATCTGGGGGAAACCTTTCGATTCACCCCCTGTATTCTGTTCAAAAGGTTCAACCCTCTTTTCGGTACGGAAT




GTTATTTTGTGAATTACTCAGTTAATTAATTTATTTTTTAAATGTGATTAACACCCAGTAACCCACTAGTCCACACAAAACCTAA世нНبН ب CAATAAAACACTTAATGAGTCAATTAATTAAATAAAAAATTTACACTAATTGTGGGTCATTGGGTGATCAGGTGTGTTTTGGATT

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GTCCTGGAGAATAATCTACGTCCAATCCTTCTCATCGAACCAGGGCAAAAACTACAAGATGGAGATATGACCCAGCATTCCATTG

CAGGACCTCTTATTAGATGCAGGTTAGGAAGAGTAGCTTGGTCCCGTTTTTGATGTTCTACCTCTATACTGGGTCGTAAGGTAAC


CTAGGAATTCATCCTAGAAAATCTCACCCAGATACCTAGGAGACACAGGCCAGAATGTCCCTGCAGCTGGAAGTGAAATTAAGGT

GATCCTTAAGTAGGATCTTTTAGAGTGGGTCTATGGATCCTCTGTGTCCGGTCTTACAGGGACGTCGACCTTCACTTTAATTCCA



TTTATTTATTTTTTTTTATTTTTTGTTTTTATTTTTTGTTTTTGAGATGGAGTCTCGTTCTGTCACCCAGGCTGGAGTGCGGTGG
 AAATAAATAAAAAAAAATAAAAAACAAAAATAAAAAACAAAAACTCTACCTCAGAGCAAGACAGTGGGTCCGACCTCACGCCACC


CGCCATCTCGGCTCACTGCAAGCTCCGCCTCCCAGGTTCACTCCATTCTCCTGCCTCAGCCTCCCGAGTAGCTGGGACTACAGGT
 GCGGTAGAGCCGAGTGACGTTCGAGGCGGAGGGTCCAAGTGAGGTAAGAGGACGGAGTCGGAGGGCTCATCGACCCTGATGTCCA


GCCCGCCATCATACCTGGCTAATTTTTTGTATTTTTAGTAGAGATGGGGTTTCACCGTGTTAGCCAGGATGGTCTTGATCTCCTG بнНبН ب CGGGCGGTAGTATGGACCGATTAAAAAACATAAAAATCATCTCTACCCCAAAGTGGCACAATCGGTCCTACCAGAACTAGAGGAC


ACCTCGTGATCCACCCGCCTTGGCCTCCCAAAGTGCTGGGATTACAGGCGTGAGCCACCACACCCAGCCAGCTTAATAATTTATA
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ATAACTGAATGTTGTACTGTTTTCTGCCATTATAGAAAATTATGTTGTTGGAGAAAACAAAATACATACAAACAAGCAAACCTTC HHץبН
TATTGACTTACAACATGACAAAAGACGGTAATATCTTTTAATACAACAACCTCTTTTGTTTTATGTATGTTTGTTCGTTTGGAAG


CCTACATAAATGACCCAAGTAGTTAAAGAATAAAACCAATTTCTTTCCATTAAAAAGAAAAGAAAGCCGGGTGTGATGCCTCATG





CCATCTCTAAGAAATAAAAGTAGGCCAGGCACAGTGGCTCACACCTATAATCCCAGCACTTTGAGAGGCGGAGGCAGGTGGATCA
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GTGGCACATGCCTGTAATCCCAGCTACTTGGGAGGCTGAGGAAGGAGAATCACTTGAACTGGGGAGGCAGAGGTTGCAGTAAGCT بץبץبץ
CACCGTGTACGGACATTAGGGTCGATGAACCCTCCGACTCCTTCCTCTTAGTGAACTTGACCCCTCCGTCTCCAACGTCATTCGA


GAGATTGCACCACTGCACTCCAGCCTGGGTGACAGAATGAGACTCCGTCTCAAAAAAAAAAAAAGAAAAATTTTAAAATGTCCTG



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CTCTACATATATATATATATATATATATATTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTGAGACAGAGTCTCACTCTAT
 GAGATGTATATATATATATATATATATATAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAACTCTGTCTCAGAGTGAGATA


TGCCCAGGCTGGAGTGCAGTGGCGCAATCTTGGCTCACTACAACCTCCACCTCCCGGGTTCAAGCCATTCTCCCGCCTCAGCCTC
 ACGGGTCCGACCTCACGTCACCGCGTTAGAACCGAGTGATGTTGGAGGTGGAGGGCCCAAGTTCGGTAAGAGGGCGGAGTCGGAG


CCAAGTAGCTGGGATTAGAGGCATGCACCACCACCCCCGGCTAATTTTGTATTTTTTGTAGAGACCGGGATTCAGCAATTTGCCC HبН GGTTCATCGACCCTAATCTCCGTACGTGGTGGTGGGGGCCGATTAAAACATAAAAAACATCTCTGGCCCTAAGTCGTTAAACGGG


AGGCTGGTCTCGAAATCCTGATCTCAGGTGATCCACCTGCCCTGGCCTCCCAAAGTGCTGGGATTACAAGCGTGAGCCACCACGC





TTAGTGGTTGCTAGGGTCTGGGTGAGGGAGAGTGGGGAGTAACTGCTCATGGGGACAGGGCCTCCTTTGGGGGTGATGAAAATGT
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TTTGGAACTTGATAGAGGTGATAGTTGCAGAATATTGTGCATGTACCTAAAGGCACTGAATTGTGTAATTCAAAGTGTGAATTTT
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ATGTTATGTGAATTTCACCTCAGTTTTTTTTAAGGTAAGAAAATGGTTATTACAAAATTCAGGATGGTAGTTATATCACAGTGTC世нНبН ب TACAATACACTTAAAGTGGAGTCAAAAAAAATTCCATTCTTTTACCAATAATGTTTTAAGTCCTACCATCAATATAGTGTCACAG


TCTGGAAACTTCCAGGGTATCCACATGTCCCTTTTTATTTTATTTTATTTTTTATTTTATTTGAGATAGGGTCTTGCTCTGTTGC HHبHبH H
AGACCTTTGAAGGTCCCATAGGTGTACAGGGAAAAATAAAATAAAATAAAAAATAAAATAAACTCTATCCCAGAACGAGACAACG


CCAGGCTAGAGTGCAGTGGCAGGATCATGACCCTCTCCTGTCTCAAATTCCTAGGCTCAAGCTATCCTCCCTCCTCAGCCTCCTA

GGTCCGATCTCACGTCACCGTCCTAGTACTGGGAGAGGACAGAGTTTAAGGATCCGAGTTCGATAGGAGGGAGGAGTCGGAGGAT


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GCCTTCTCCTAATTGTTGACATAGGTAGTAGTTGCATGACATTCACTTTGTAATTATGTGTTTCAGGAATTCTCAGGCCTGTGGG
 CGGAAGAGGATTAACAACTGTATCCATCATCAACGTACTGTAAGTGAAACATTAATACACAAAGTCCTTAAGAGTCCGGACACCC


AGCTCTTAATAAATAAAAAAGAGGCCAGGTGTGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGAGGCAGGCGGATCA
 TCGAGAATTATTTATTTTTTCTCCGGTCCACACCACCGAGTGCGGACATTAGGGTCGTGAAACCCTCCGGCTCCGTCCGCCTAGT



GCGGGTGCCTGTAATCCCAGTTACTTGGGAGGCTGAGGCAGGAGAATCGCTTGAACCTGGGAGGCGGAGGTTGCAGTAAGCTGAG HبН CGCCCACGGACATTAGGGTCAATGAACCCTCCGACTCCGTCCTCTTAGCGAACTTGGACCCTCCGCCTCCAACGTCATTCGACTC


ATCGCGCCACTGCACACCAGCCTGGGTGATAAGAGCAAGACTCCATCTCAAAATAAATGAATAAATAAAAATAAATAAATAAATA

TAGCGCGGTGACGTGTGGTCGGACCCACTATTCTCGTTCTGAGGTAGAGTTTTATTTACTTATTTATTTTTATTTATTTATTTAT
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PCR Forward
actgcactccagcctgggtgacacagtgagacccgatctctatagatanatgantggatgantgaggaggtcanggatcctcacc
 tgacgtgaggtcgancccactgtatcactctggaccagagatatctatttacttacctacttactcccccagttcctaggagtag


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\frac{\text { Sanger Sequencing }}{\text { AATGCTTGCCAGTTCTGGAG }}
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CGGCTTCCATTTGGAGGGAGGAGTTTGGTTGAGTTCTTGCAAGGTTGGTACCTAGGAAATGCTTGCCAGTTCTGGAGCCCAGACA
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ACATTTGTACAGGGGGTGGGGATGGGGGACATGGTGGGGCCGCCTCCAGAAAGTTGGGAAAGTGAGCCTCGTGTTTCGAGGGCTG
 TGTAAACATGTCCCCCACCCCTACCCCCTGTACCACCCCGGCGGAGGTCTTTCAACCCTTTCACTCGGAGCACAAAGCTCCCGAC CACNA1A


Donor Template WT -> SNV
TGTAAACATGTC
Donor Template WT -> SNV


CGTCTCTCTCCTCAACTCCATGAAGTCCATCATCAGCCTGTTGTTTCTCCTTTTCCTGTTCATTGTCGTCTTCGCCCTTTTGGGA
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ATGCAACTCTTCGGCGGCCAGTAAGTCCTTCACAGGAATTCCAACTCCTGGTTCCCTGGGGTCAGGCTCAGGGAACACACAGTCC
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TTTCCTGGGGACAGAGATGATGGAGACGTTCGTTTCCTTGGAGATGAGATACTGAGCCACACCCTCAGAGCACCCCGGGTGGGGC
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CAACGTGAAATGTCTGTGTCCTCCCTGCAGGTTTAATTTCGATGAAGGGACTCCTCCCACCAACTTCGATACTTTTCCAGCAGCA
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 TATTACTGCCACAAAGTCCATGTCGGAGGTGGACCGGGGTGCCCGGTTGTGGAGAGTCACAGTGTCTACTTTCACGGACGAGGTG



GCCACAAAGTCCATGTCGGAGGTGG
P PCR Reverse

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| $\square$ CACNA1A |
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|  |  |  | 895 |  |  |  |  | 00 |  |  |  |  | 905 |  |  |  |  | 10 |  |  |  |  | 15 |  |  |  |  |
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| S | R | E | G | P | Y | G | R | E | S | D | H | H | A | R | E | G | S | L | E | Q | P | G | F | W | E | G | E |
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20,614
$5^{\prime}$

| CACNA1A |  |
| :--- | :--- |
| /note $\quad=$ | gene ENSG000000141837 <br>  <br> Protein coding |

CACNA1A-201
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| CACNA1A-254 |  |
| ---: | :--- |
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| CACNA1A-232 |  |
| :--- | :--- |
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[^0]Feature
Location
Size
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/translation = SPFARASIKSAKLENSTFFHKKERRMRFYIRRMVKTQAFYWTVLSLVALNTLCVAIVHYNQPEWLSDFL,,YYAEFIFLGLFMSEMFIKMYGLGTRP YFHSSFNCFDCG,,VIIGSIFEVIWAVIKPGTSFGISVLRALRLLRIFKVTK,,YWASLRNLVVSLLNSMKSIISLLFLLFLFIVVFALLGMQLFGGQ,,FN FDEGTPPTNFDTFPAAIMTVFQ,,ILTGEDWNEVMYDGIKSQGGVQGGMVFSIYFIVLTLFGN,,YTLLNVFLAIAVDNLANAQELTK,,DEQEEEEA ANQ KLA LQKAKEVAEVSPLSAANMSIAV,,KEQQKNQKPAKSV WEQRTSEMRKQNLLASREALYNEMDPDERWKAAYTRHLRPDMKTHLDRPL VVDPQENRNNNTNKSRAAEPTVDQRLGQQRAEDFLRKQARYHDRARDPSGSA GLDARRPWAGSQEAELSREGPYGRESDHHAREGSLEQPG FWE GEAERGKA GDPHRRHVHRQ GGSRESRSGSPRTGADGEHRRHRAHRRPGEEGPEDKAERRARHREGSRPARGGEGEGEGPDGGERRRRHR





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1614 .. 20,391 18,778 bp $\quad \rightarrow \quad \mathrm{CDS}$

* 9 segments $=1744 \mathrm{bp}$
/codon_start = 1
/note $\quad=$ coding sequence ENSP00000490578
/translation $=$
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/note $\quad=$ coding sequence ENSP00000489777
/translation $=$
SPFARASIKSAKLENSTFFHKKERRMRFYIRRMVKTQAFYWTVLSLVALNTLCVAIVHYNQPEWLSDFL,,YYAEFIFLGLFMSEMFIKMYGLGTRP YFHSSFNCFDCG,,VIIGSIFEVIWAVIKPGTSFGISVLRALRLLRIFKVTK,,YWASLRNLVVSLLNSMKSIISLLFLLFLFIVVFALLGMQLFGGQ,, FN FDEGTPPTNFDTFPAAIMTVFQ,,ILTGEDWNEVMYDGIKSQGGVQGGMVFSIYFIVLTLFGN,,YTLLNVFLAIAVDNLANAQELTK,,DEQEEEEA ANQKLALQKAKEVAEVSPLSAANMSIAV,,KEQQKNQKPAKSVWEQRTSEMRKQNLLASREALYNEMDPDERWKAAYTRHLRPDMKTHLDRPL VVDPQENRNNNTNKSRAAEPTVDQRLGQQRAEDFLRKQARYHDRARDPSGSA GLDARRPWAGSQEAELSREGPYGRESDHHAREGSLEQPG FWEGEAERGKA GDPHRRHVHRQGGSRESRSGSPRTGADGEHRRHRAHRRPGEEGPEDKAERRARHREGSRPARGGEGEGEGPDGGERRRRHR

/translation = SPFARASIKSAKLENSTFFHKKERRMRFYIRRMVKTQAFYWTVLSLVALNTLCVAIVHYNQPEWLSDFL,,YYAEFIFLGLFMSEMFIKMYGLGTRP YFHSSFNCFDCG,,VIIGSIFEVIWAVIKPGTSFGISVLRALRLLRIFKVTK,,YWASLRNLVVSLLNSMKSIISLLFLLFLFIVVFALLGMQLFGGQ,,FN FDEGTPPTNFDTFPAAIMTVFQ,,ILTGEDWNEVMYDGIKSQGGVQGGMVFSIYFIVLTLFGN,,YTLLNVFLAIAVDNLANAQELTKVEA ,,DEQEE EEAANQKLALQKAKEVAEVSPLSAANMSIAV,,KEQQKNQKPAKSV WEQRTSEMRKQNLLASREALYNEMDPDERWKAAYTRHLRPDMKTHLD RPLVVDPQENRNNNTNKSRAAEPTVDQRLGQQRAEDFLRKQARYHDRARDPSGSA GLDARRPWAGSQEAELSREGPYGRESDHHAREGSLE QPGFWEGEAERGKA GDPHRRHVHRQGGSRESRSGSPRTGADGEHRRHRAHRRPGEEGPEDKAERRARHREGSRPARGGEGEGEGPDGGERR


CACNA1A-245

* 9 segments $=1744 \mathrm{bp}$


## /codon_start = 1

/note $\quad=$ coding sequence ENSP00000489861
/translation $=$
SPFARASIKSAKLENSTFFHKKERRMRFYIRRMVKTQAFYWTVLSLVALNTLCVAIVHYNQPEWLSDFL,,YYAEFIFLGLFMSEMFIKMYGLGTRP YFHSSFNCFDCG,,VIIGSIFEVIWAVIKPGTSFGISVLRALRLLRIFKVTK,,YWASLRNLVVSLLNSMKSIISLLFLLFLFIVVFALLGMQLFGGQ,,FN FDEGTPPTNFDTFPAAIMTVFQ,,ILTGEDWNEVMYDGIKSQGGVQGGMVFSIYFIVLTLFGN,,YTLLNVFLAIAVDNLANAQELTK,,DEQEEEEA ANQ KLA LQ KA KEVAEVSPLSAANMSIAV,,KEQ QKNQ KPAKSV WEQRTSEMRKQNLLA SREA LYNEMDPDERWKAAYTRHLRPDMKTHLDRPL VVDPQENRNNNTNKSRAAEPTVDQRLGQQRAEDFLRKQARYHDRARDPSGSA GLDARRPWAGSQEAELSREGPYGRESDHHAREGSLEQPG FWEGEAERGKA GDPHRRHVHRQGGSRESRSGSPRTGADGEHRRHRAHRRPGEEGPEDKAERRARHREGSRPARGGEGEGEGPDGGERRRRHR


$$
1614 \text {.. 20,391 18,778 bp } \quad \square \quad \rightarrow \quad \text { CDS }
$$

## CACNA1A-246

1614 .. 20,391 18,778 bp $\quad \rightarrow \quad \mathrm{CDS}$

* 9 segments $=1744 \mathrm{bp}$
/codon_start $=1$
/note $\quad=$ coding sequence ENSP00000489778
$\begin{aligned} \text { /translation }= & \text { SPFARASIKSAKLENSTFFHKKERRM } \\ & \text { YFHSSFNCFDCG,,VIIGSIFEVIWAV } \\ & \text { FDEGTPPTNFDTFPAAIMTVFQ,,ILT } \\ & \text { ANQKLALQKAKEVAEVSPLSAANM } \\ & \text { VVDPQENRNNNTNKSRAAEPTVDQ } \\ & \text { FWEGEAERGKA GDPHRRHVHRQGG } \\ & \text { H\&ARATHKEGEQARREGKK. }\end{aligned}$


CACNA1A-252
1614 .. 20,391 18,778 bp $\quad \rightarrow \quad \mathrm{CDS}$

* 9 segments $=1744 \mathrm{bp}$
/codon_start = 1
/note $\quad=$ coding sequence ENSP00000489715
/translation =
SPFARASIKSAKLENSTFFHKKERRMRFYIRRMVKTQAFYWTVLSLVALNTLCVAIVHYNQPEWLSDFL,,YYAEFIFLGLFMSEMFIKMYGLGTRP YFHSSFNCFDCG,,VIIGSIFEVIWAVIKPGTSFGISVLRALRLLRIFKVTK,,YWASLRNLVVSLLNSMKSIISLLFLLFLFIVVFALLGMQLFGGQ,,FN FDEGTPPTNFDTFPAAIMTVFQ,,ILTGEDWNEVMYDGIKSQGGVQGGMVFSIYFIVLTLFGN,,YTLLNVFLAIAVDNLANAQELTK,,DEQEEEEA A NQ KLA LQ KA KEVAEVSPLSAANMSIAV,,KEQ Q KNQ KPAKSV WEQRTSEMRKQNLLA SREA LYNEMDPDERWKAAYTRHLRPDMKTHLDRPL VVDPQENRNNNTNKSRAAEPTVDQRLGQQRAEDFLRKQARYHDRARDPSGSA GLDARRPWAGSQEAELSREGPYGRESDHHAREGSLEQPG FWE GEA ERGKA GDPHRRHVHRQ GGSRESRSGSPRTGADGEHRRHRAHRRPGEEGPEDKAERRARHREGSRPARGGEGEGEGPDGGERRRRHR

CACNA1A-256
1614 .. 20,391 18,778 bp $\quad \rightarrow \quad$ CDS
* 9 segments $=1744 \mathrm{bp}$
/codon_start = 1
/note $\quad=$ coding sequence ENSP00000489913
/translation $=$
SPFARASIKSAKLENSTFFHKKERRMRFYIRRMVKTQAFYWTVLSLVALNTLCVAIVHYNQPEWLSDFL,,YYAEFIFLGLFMSEMFIKMYGLGTRP YFHSSFNCFDCG,,VIIGSIFEVIWAVIKPGTSFGISVLRALRLLRIFKVTK,,YWASLRNLVVSLLNSMKSIISLLFLLFLFIVVFALLGMQLFGGQ,,FN FDEGTPPTNFDTFPAAIMTVFQ,,ILTGEDWNEVMYDGIKSQGGVQGGMVFSIYFIVLTLFGN,,YTLLNVFLAIAVDNLANAQELTK,,DEQEEEEA A NQ KLA LQ KA KEVAEVSPLSAANMSIAV,,KEQQ KNQ KPAKSVWEQRTSEMRKQNLLA SREA LYNEMDPDERWKAAYTRHLRPDMKTHLDRPL VVDPQENRNNNTNKSRAAEPTVDQRLGQQRAEDFLRKQARYHDRARDPSGSA GLDARRPWAGSQEAELSREGPYGRESDHHAREGSLEQPG FWE GEA ERGKA GDPHRRHVHRQ GGSRESRSGSPRTGADGEHRRHRAHRRPGEEGPEDKAERRARHREGSRPARGGEGEGEGPDGGERRRRHR


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9 segments = 1753 bp
```

/codon_start = 1
/note $\quad=$ coding sequence ENSP00000489829
/translation $=$ SPFARASIKSAKLENSTFFHKKERRMRFYIRRMVKTQAFYWTVLSLVALNTLCVAIVHYNQPEWLSDFL,,YYAEFIFLGLFMSEMFIKMYGLGTRP
YFHSSFNCFDCG,,VIIGSIFEVIWAVIKPGTSFGISVLRALRLLRIFKVTK,,YWASLRNLVVSLLNSMKSIISLLFLLFLFIVVFALLGMQLFGGQ,,FN
FDEGTPPTNFDTFPAAIMTVFQ,,ILTGEDWNEVMYDGIKSQGGVQGGMVFSIYFIVLTLFGN,,YTLLNVFLAIAVDNLANAQELTKVEA,,DEQEE
EEAANQKLALQKAKEVAEVSPLSAANMSIAV,,KEQQKNQKPAKSVWEQRTSEMRKQNLLASREALYNEMDPDERWKAAYTRHLRPDMKTHLD
RPLVVDPQENRNNNTNKSRAAEPTVDQRLGQQRAEDFLRKQARYHDRARDPSGSA GLDARRPWAGSQEAELSREGPYGRESDHHAREGSLE
QPGFWEGEAERGKA GDPHRRHVHRQ GGSRESRSGSPRTGADGEHRRHRAHRRPGEEGPEDKAERRARHREGSRPARGGEGEGEGPDGGERR




/note $\quad=\quad$| primary transcript ENST000000637117 |
| :--- |
|  |
| Retained intron |

| Donor Template WT -> SNV | 10,453 | .. | 10,552 | 100 bp | $\square$ | $\mapsto$ |
| :--- | ---: | ---: | ---: | ---: | ---: | :--- | misc_feature

    \(=W T=G\)
    \(S N V=A\)
    PAM
10,491 .. $10,492 \mathrm{bp} \quad \square \quad$ misc_feature



[^0]:    * 9 segments $=1744 \mathrm{bp}$
    /codon_start = 1
    /note $\quad=$ coding sequence ENSP00000353362
    /translation = SPFARASIKSAKLENSTFFHKKERRMRFYIRRMVKTQAFYWTVLSLVALNTLCVAIVHYNQPEWLSDFL,,YYAEFIFLGLFMSEMFIKMYGLGTRP YFHSSFNCFDCG,,VIIGSIFEVIWAVIKPGTSFGISVLRALRLLRIFKVTK,,YWASLRNLVVSLLNSMKSIISLLFLLFLFIVVFALLGMQLFGGQ,,FN FDEGTPPTNFDTFPAAIMTVFQ,,ILTGEDWNEVMYDGIKSQGGVQGGMVFSIYFIVLTLFGN,,YTLLNVFLAIAVDNLANAQELTK,,DEQEEEEA A NQ KLA LQ KA KEVAEVSPLSAANMSIAV,,KEQQ KNQ KPAKSVWEQRTSEMRKQNLLA SREA LYNEMDPDERWKAAYTRHLRPDMKTHLDRPL VVDPQENRNNNTNKSRAAEPTVDQRLGQQRAEDFLRKQARYHDRARDPSGSA GLDARRPWAGSQEAELSREGPYGRESDHHAREGSLEQPG FWE GEA ERGKA GDPHRRHVHRQ GGSRESRSGSPRTGADGEHRRHRAHRRPGEEGPEDKAERRARHREGSRPARGGEGEGEGPDGGERRRRHR
    

