

INK2J00028.1_PSEN1_E276A_A03.2_BB
26,923 bp

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GATATTAATATCTAATGTTTGGGAGCCATCACATTATTCTAAATAATGTTTTGGTGAAAATTATTGTACATCTTTTAAAATCTGT
 CTATAATTATAGATTACAAACCCTCGGTAGTGTAATAAGATTTATTACAAAACCACTTTTAATAACATGTAGAAAATTTTAGACA


GTAATTTTTTTTCAGGGAAGTGTTTAAAACCTATAACGTTGCTGTGGACTACATTACTGTTGCACTCCTGATCTGGAATTTTGGT ب- ب
CATTAAAAAAAAGTCCCTTCACAAATTTTGGATATTGCAACGACACCTGATGTAATGACAACGTGAGGACTAGACCTTAAAACCA


GTGGTGGGAATGATTTCCATTCACTGGAAAGGTCCACTTCGACTCCAGCAGGCATATCTCATTATGATTAGTGCCCTCATGGCCC HبНبН ب CACCACCCTTACTAAAGGTAAGTGACCTTTCCAGGTGAAGCTGAGGTCGTCCGTATAGAGTAATACTAATCACGGGAGTACCGGG


TGGTGTTTATCAAGTACCTCCCTGAATGGACTGCGTGGCTCATCTTGGCTGTGATTTCAGTATATGGTAAAACCCAAGACTGATA
 ACCACAAATAGTTCATGGAGGGACTTACCTGACGCACCGAGTAGAACCGACACTAAAGTCATATACCATTTTGGGTTCTGACTAT



TCCCATAACTCTTCAGTAAATCATTAATTAGCTATAGTAACTTTTTCATTTGAAGATTTCGGCTGGGCATGGTAGCTCATGCCTG
 AGGGTATTGAGAAGTCATTTAGTAATTAATCGATATCATTGAAAAAGTAAACTTCTAAAGCCGACCCGTACCATCGAGTACGGAC

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AATTAGCTGGGTGTGGTGGCGTACCCTTGTATTCCCAGCTACTTGAGAGGCTGAGGCAGGAGAATTATTTGAACCCAGGAAGTGA
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CTACTTTTTATAAGGTAATGTATAGAATTATTCTTTTAAAAATAAAACCGATTTGGACAGTGTGAGATTCACATTCTGTAACCAC ب+ GATGAAAAATATTCCATTACATATCTTAATAAGAAAATTTTTATTTTGGCTAAACCTGTCACACTCTAAGTGTAAGACATTGGTG


CAGTGTGACATGGGTCCTGAACAGTTAGAACATACTCCAGCCATTAACCCAGGCAGCTTTCAGGTACGTACTCTGTGGCTGTTGC
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GATCCTCCCACCTTGGCCTGCCAAAGTGTTGGGATTACAGGCATGAGCCACCATGCCTGGCCATACACTTTTTTTTTTTTTTTTT
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CATCTATTCTTGCTTAGCTGTGAGTAAGAATGCCAGGTCTGGAGACAGAATGTCTGGGTTCAAATTCTACTCATCACTTTTTATT بнНبץبץبץ GTAGATAAGAACGAATCGACACTCATTCTTACGGTCCAGACCTCTGTCTTACAGACCCAAGTTTAAGATGAGTAGTGAAAAATAA


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CCGGGATCAAGCGATTCTCCTGCCTCAGCCTTCCGAGTAGCTGGGACTACAAGTGCGCACCACCACGCCCAGCTAATTTTTGTAT
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CCAAAGTGCTGAGATTACAGGTGTGAGCCACTATACCCAGCCCAGTGTTATATTTTTGTATAATCCTATGAAGTATCAAGGCAGT
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TGTGTTTTATTACTGTACTATATTATATTATAGTTCTATATATACTATATTATATTGTACATATATATTATTATATGAAGAGACA ب+ ACACAAAATAATGACATGATATAATATAATATCAAGATATATATGATATAATATAACATGTATATATAATAATATACTTCTCTGT


TGACCACTTTGAGGCATGAAATTTTTTTTTTTTTTTTTTTTTTTTTGAGGCACGGTCTTACTTTGTCACCTAGGCAGTGGCGCCA

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TCTTGGCTCACTGCAGCCTCCACCTCCAGGGCTCAAGCAATCCTCCCACCTCAGCTTCCTGAGTAGCTGGGACTATAGGCACCTG



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AGTGTTTAAAATTATTTTTATATTTTTATATTCTGAATTTTCTTTCTTTCTTTCTTTTTTTTTTTTTTTTTGAGACGGAGTCTC
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CAGCCTCCCGAGTAGCTGGGACTACAGGCACGCGCCACCACACCCAGCTAATTTTTTGTATTTTTTAGTAGAGACAGGGTTTCAC

GTCGGAGGGCTCATCGACCCTGATGTCCGTGCGCGGTGGTGTGGGTCGATTAAAAAACATAAAAAATCATCTCTGTCCCAAAGTG


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GCACAATCGGTCCTACCAGAACTAGAGGACTGGAACACTAGGCGGGTGGAGCCGGAGGGTTTCACGACCCTAACGTCCGCACTCG





GCCCTCTTACTTCAAGTACTTCTCAGTAGTACGTAATACTGCTTTTAAAAAGGAGAAGGGATGAATTTCTTCTACTCTGCTCTTC بнНبНبץب+ CGGGAGAATGAAGTTCATGAAGAGTCATCATGCATTATGACGAAAATTTTTCCTCTTCCCTACTTAAAGAAGATGAGACGAGAAG


ATATTTTGAAAAGTTCAGTCAAATCCCCTTTATTAAATTCATCTCAGAGTAATCTTTTTAATTTGTAGTTCATATCCGTGATTAG بम+Н $+\boldsymbol{+}$ TATAAAACTTTTCAAGTCAGTTTAGGGGAAATAATTTAAGTAGAGTCTCATTAGAAAAATTAAACATCAAGTATAGGCACTAATC


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| TTCTCCCTGTTTCTGCTCACTGTAG |

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gRNA Protospacer
T
TAGCCCATACATTTTATTAGATGTCTTTTATGTTTTTCTTTTTCTAGATTTAGTGGCTGTTTTGTGTCCGAAAGGTCCACTTCGT
 ATCGGGTATGTAAAATAATCTACAGAAAATACAAAAAGAAAAAGATCTAAATCACCGACAAAACACAGGCTTTCCAGGTGAAGCA


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AGGCTTTCCAGGTGAAGCA
Donor Template WT -> SNV




TATGAATAGAAAGAAAGAAAATGTTTAGATATTGGGGAACCAGCATTCCCATTTTAAAACCTGTTAGGAGTTGTTGATTAGGGCA
 ATACTTATCTTTCTTTCTTTTACAAATCTATAACCCCTTGGTCGTAAGGGTAAAATTTTGGACAATCCTCAACAACTAATCCCGT
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 TCGAGTTCCTAAGGAAACTCACTGACCAAATCTACAGAAAGACGATAAGCCACTGGTGACCCCTTGACTCTAACAACTCGTCTTC


GGTAATGTGAGCAGAGCCGTGCCTTTGTAAGCTGGCAGCACTGTGTGAGATGAATTGGTGGGTTGGATACTGAGATCATGAGAGG НННННННННННبН CCATTACACTCGTCTCGGCACGGAAACATTCGACCGTCGTGACACACTCTACTTAACCACCCAACCTATGACTCTAGTACTCTCC

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CATACTAAGCATAATTAAGATGATATTGCCATGATCTAGGTGGAAAGTAATGGGGGTTTGAATTATGGTAGTGGCAGTAGCAATC HبН





ACGCACTACCAGGCCTGGCTAATTTTTTTGTGTTGTGTGTAGAGACTGGGTTTTGCCATGTGCCCAGGCTGGTCTTGAACTCCTC
 TGCGTGATGGTCCGGACCGATTAAAAAAACACAACACACATCTCTGACCCAAAACGGTACACGGGTCCGACCAGAACTTGAGGAG


GGCTTAAGCGATCCTCCTGCCTTGACTTCACAAAGTGCTTGAGTTACAGGTGTGAGCTACCACGCCTGGCCATGTTTTCTTGTGT
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TCATGTGTTTCTTAGTTACAGATCTGAATTTATTTTGTAACTGGCTTGGTATAATCTTTTTCATATTTGTGAAATTAATCTTTTT ب+ AGTACACAAAGAATCAATGTCTAGACTTAAATAAAACATTGACCGAACCATATTAGAAAAAGTATAAACACTTTAATTAGAAAAA


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GAGGATCACTTGAACCCAGGAGGCGGAGGTTACAGTGAGCCATGATCGTGCCACTGCACTCCAGCCTGAGTGACAGCAAGGCTTC





AGCAGCTGGGACTACAGGCACACGCCGCCACGCCCGGCTAATTTTTGTATTTTTAGTAGCAACGGGGTTTCACCATGTTAGCCAG بн十ب中 TCGTCGACCCTGATGTCCGTGTGCGGCGGTGCGGGCCGATTAAAAACATAAAAATCATCGTTGCCCCAAAGTGGTACAATCGGTC


GATGGTCTCGATCTCCTGACCTTGTGATCCGCCCGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGCGTGAGCCACCGCGCCTGG بнبץب CTACCAGAGCTAGAGGACTGGAACACTAGGCGGGCGGAGCCGGAGGGTTTCACGACCCTAATGTCCGCACTCGGTGGCGCGGACC


GTGACATTGAAGAGAGCAGTTTCAGTAGAGTGATAGAACCAGAAGCAGTACTCCAAAGGGAAGGTGCGTATGTGTGCATGCATGT

CACTGTAACTTCTCTCGTCAAAGTCATCTCACTATCTTGGTCTTCGTCATGAGGTTTCCCTTCCACGCATACACACGTACGTACA


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GAGGTAAGTCTTGCTCTGTTGCCCAGGCTGGAGTGCAGTGGCGCAATCTCAGCTCACTGCAACCTCCATCTCCTGGGTTCAGGCA بץبНبץب+
CTCCATTCAGAACGAGACAACGGGTCCGACCTCACGTCACCGCGTTAGAGTCGAGTGACGTTGGAGGTAGAGGACCCAAGTCCGT


ATTCTCCTGCCTCAGCCTCCCGAGTAGTTGAGATTACAAGCATGCGCCACTATACCTGGCTAATTTTTTTTTCCATATTTTTAG

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GGGTTAGCAAACTACTGCTTGAAGCCAAATATGGCCCATGGCTTGTTTTTGTACAGCTTATAAGCTAAAAATGCTTTTTACATTT
 CCCAATCGTTTGATGACGAACTTCGGTTTATACCGGGTACCGAACAAAAACATGTCGAATATTCGATTTTTACGAAAAATGTAAA


AAAAAAAAAAAAAAAGAACAAGGAAGAATATGTGACACACAAAACCTGAAATGTTTACTCCCTGGCCATTTACAGAAAAAGTTAG

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AACCTAATAAATCGCTGGTGGCTTCATTTCCAAAAGGCTATGGAAATCCATAAACAGGCTGGGTGCAGTGGCTCATGCCTGTAAT
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CAGAAATAACAAGGTGGTCGTTCAGCTTCTTTGATTGTGTGTTTCTTTCATAAGTTCTTCAAGAAGCCTGAGTATTAGAAACATG بץبнبץ
GTCTTTATTGTTCCACCAGCAAGTCGAAGAAACTAACACACAAAGAAAGTATTCAAGAAGTTCTTCGGACTCATAATCTTTGTAC


GATAAAATTATTGTGATAAAAAGCCAGAGAGACATAAATGTCAAGTATCTTGTTTAAAATTACTGTGACCAGGCCGGGCGTGGTG
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 ACTCCGTCCTCTTACCGCACTTGGGTCCTCTGCCTCGAACGTCACTCGGCTCTAGTACGGTGACATGAGGTCGGACCCACTGTCT

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GCAAGACTCTGTCTCAAAAAAAAAAAAAAAAAAAAAAATTACTGTGACCAGATTGGACTCAGCTTGCTGCTGGCATTTGGTTCCC
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ACCATAACCTCATATGTCATGTGTTTGCTTATATGTACTTTTGTGTTATTGTTGGTGTATCTTCAGGATAAGTTCCAAAATGTAA + $+\boldsymbol{+}$ TGGTATTGGAGTATACAGTACACAAACGAATATACATGAAAACACAATAACAACCACATAGAAGTCCTATTCAAGGTTTTACATT


TATTGCTGGGTTAAAGGATTAATGCACATGTAGTTTTATTAGATGTTACCAAATTTCCCTCCAGTGGGGATTATACCATTTTTCA

ATAACGACCCAATTTCCTAATTACGTGTACATCAAAATAATCTACAATGGTTTAAAGGGAGGTCACCCCTAATATGGTAAAAAGT


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 TACTTCTCAAAACACTATCGTCCACGTCAAACTCATGATGTCATTTGTAAGTCAAAGTCTTGAAGAAGAGAATGGACGATTTTGG


AAAGAGAACCTTTTTTTTATTTTTACTTCTGATTGTTGAACAGTCTTAAGGCAGCATTAGGAAGACTGGCGATTTGTGTGGAGAA

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TTATTAGATAATATCTTGATTTTTCAGGGTCACTGTTATAAGCTAACAGTATAGCAATGTTTTTATCGTCTTTCTTTGGTCATAG ب+ AATAATCTATTATAGAACTAAAAAGTCCCAGTGACAATATTCGATTGTCATATCGTTACAAAAATAGCAGAAAGAAACCAGTATC


ACTCCTTTGAGAATCTCTTGAGAACTATGATAATGCCCAGTAAATACACAGATAAGTATTTAAGGAGTTCAGATACTCAAAACCC
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CACTTTCTCTTGAAGGCACAGAAAGGGAGTCACAAGACACTGTTGCAGAGAATGATGATGGCGGGTTCAGTGAGGAATGGGAAGC + + ب $+\boldsymbol{+}+\boldsymbol{+} \boldsymbol{+}$ gtganagagancttccgtgtctttccctcagtgttctgtgacancgtctcttactactaccgcccaagtcactccttacccttcg


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| :---: |
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$3^{\prime}$
26,923 $5^{\prime}$

| Featur |  |  | Locati | ation | Size | $\square$ | 7 | Type |
| :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: |
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| /note | $\begin{aligned} = & \text { gene ENSG00000080815 } \\ & \text { Protein coding } \end{aligned}$ |  |  |  |  |  |  |  |
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| /note | $=$ primary transcript ENST00000556951 |  |  |  |  |  |  |  |
| PSEN1-220 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000557293 |  |  |  |  |  |  |  |
| PSEN1-223 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000559361 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |  |
| PSEN1-225 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENSTO00000697912 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |  |
| PSEN1-226 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | ```= primary transcript ENST00000697913 Retained intron``` |  |  |  |  |  |  |  |
| PSEN1-229 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700265 |  |  |  |  |  |  |  |
| PSEN1-230 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST00000700266 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |  |
| PSEN1-231 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700267 |  |  |  |  |  |  |  |
| PSEN1-232 |  |  | .. | 26,923 | 26,923 bp | - | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700268 |  |  |  |  |  |  |  |
| PSEN1-233 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700269 |  |  |  |  |  |  |  |
| PSEN1-235 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700271 |  |  |  |  |  |  |  |
| PSEN1-236 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700272 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |  |
| PSEN1-237 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700273 |  |  |  |  |  |  |  |
| PSEN1-238 |  |  | .. | 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700302 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |  |


| eatur |  |  | Locatio | Size | $\square$ | \# | Type |
| :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: |
| PSEN1-239 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700303 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-240 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENSTO0000700304 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-241 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENSTO0000700305 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-242 |  |  | 1 .. 26,923 | 26,923 bp | - | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700306 |  |  |  |  |  |  |
| PSEN1-243 |  |  | 1 .. 26,923 | 26,923 bp | - | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700307 |  |  |  |  |  |  |
| PSEN1-244 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700308 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-245 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENSTO0000700309 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-246 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700310 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-247 |  |  | 1 .. 26,923 | 26,923 bp | ㅁ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST00000700311 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-248 |  |  | 1 .. 26,923 | 26,923 bp | ㅁ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700312 |  |  |  |  |  |  |
| PSEN1-249 |  |  | 1 .. 26,923 | 26,923 bp | - | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700313 |  |  |  |  |  |  |
| PSEN1-250 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST00000700314 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-251 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700315 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-252 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700316 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-253 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700317 |  |  |  |  |  |  |
| PSEN1-254 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700318 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-255 |  |  | 1 .. 26,923 | 26,923 bp | - | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENSTO0000700319 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-256 |  |  | 1 .. 26,923 | 26,923 bp | - | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700320 |  |  |  |  |  |  |
| PSEN1-257 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700321 |  |  |  |  |  |  |
| PSEN1-258 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700322 |  |  |  |  |  |  |
| PSEN1-259 |  |  | 1 .. 26,923 | 26,923 bp | - | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700323 |  |  |  |  |  |  |


| eatu |  |  | Location | Siz | $\square$ | \# | Type |
| :---: | :---: | :---: | :---: | :---: | :---: | :---: | :---: |
| PSEN1-260 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700324 |  |  |  |  |  |  |
| PSEN1-262 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700375 |  |  |  |  |  |  |
| PSEN1-264 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700377 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-265 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700378 |  |  |  |  |  |  |
| PSEN1-266 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700379 <br> protein_coding_CDS_not_defined |  |  |  |  |  |  |
| PSEN1-268 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700389 |  |  |  |  |  |  |
| PSEN1-269 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700390 } \\ & \text { Retained intron } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-271 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700404 } \\ & \text { Retained intron } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-284 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700436 <br> Nonsense mediated decay |  |  |  |  |  |  |
| PSEN1-285 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | = primary transcript ENST00000700437 |  |  |  |  |  |  |
| PSEN1-287 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700468 |  |  |  |  |  |  |
| PSEN1-288 |  |  | 1 .. 26,923 | 26,923 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700469 |  |  |  |  |  |  |
| PSEN1-212 |  |  | 1 .. 26,831 | 26,831 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENSTOOOOO } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-222 |  |  | 1 .. 26,831 | 26,831 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000557511 |  |  |  |  |  |  |
| PSEN1-208 |  |  | 1 .. 24,700 | 24,700 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000553855 } \\ & \text { Nonsense mediated decay } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-283 |  |  | 1 .. 20,965 | 20,965 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $=$ primary transcript ENST00000700435 protein_coding_CDS_not_defined |  |  |  |  |  |  |
| PSEN1-286 |  |  | 1 .. 14,879 | 14,879 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700467 } \\ & \text { Retained intron } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-234 |  |  | 1 .. 14,797 | 14,797 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700270 } \\ & \text { Retained intron } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-282 |  |  | 1 .. 14,426 | $14,426 \mathrm{bp}$ | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700434 } \\ & \text { Retained intron } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-281 |  |  | 1 .. 14,392 | 14,392 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700433 } \\ & \text { Retained intron } \end{aligned}$ |  |  |  |  |  |  |
| PSEN1-280 |  |  | 1 .. 1121 | 1121 bp | $\square$ | $\rightarrow$ | prim_transcript |
| /note | $\begin{aligned} = & \text { primary transcript ENST000000700432 } \\ & \text { Retained intron } \end{aligned}$ |  |  |  |  |  |  |


| PSEN1-263 |  |
| :--- | :--- |
| /note | $=$primary transcript ENST0000007000376 <br> Retained intron |


| PSEN1-267 |  |
| :--- | :--- |
| /note | $=$primary transcript ENST000000700388 <br>  <br> Retained intron |


| PSEN1-201 | 186 .. 26,831 | 26,646 bp | $\square \quad \rightarrow$ |  | CDS |
| :---: | :---: | :---: | :---: | :---: | :---: |
| * 6 segments | 856 bp |  |  |  |  |
| /codon_start | 1 |  |  |  |  |
| /note | coding sequence ENSP00000326366 |  |  |  |  |
| /translation | EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,DLVAVLCPKGPLRMLVETAQE RNETLFPALIYS,,STMVWLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ 2 $£ 4^{*}$ amino acids $=31.7 \mathrm{kDa}$ |  |  |  |  |
| PSEN1-202 | 186 .. 26,831 | 26,646 bp | $\square$ | $\rightarrow$ | CDS |
| * 6 segments $=856 \mathrm{bp}$ |  |  |  |  |  |
| /codon_start = 1 |  |  |  |  |  |
| /note | coding sequence ENSP00000350342 |  |  |  |  |
| /translation | EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQ | LIMIS |  |  |  | RNETLFPALIYS,,STMV WLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ 2 $844^{*}$ amino acids $=31.7 \mathrm{kDa}$



## PSEN1-207

186 .. 26,831
$26,646 \mathrm{bp}$
CDS

- 6 segments $=856 \mathrm{bp}$
/codon_start = 1
/note $\quad=$ coding sequence ENSP00000452477
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,DLVAVLCPKGPLRMLVETAQE RNETLFPALIYS,,STMVWLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ £ $844^{*}$ amino acids $=31.7 \mathrm{kDa}$

```
PSEN1-209
* 6 segments = 856 bp
/codon_start = 1
/note = coding sequence ENSP00000451915
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTAWLILAVISVY,,DLVAVLCPKGPLRMLVETAQE
    RNETLFPALIYS,,STMV WLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI
    LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ
    2%4*amino acids = 31.7 kDa
```



Feature
Location
Size
Type
/note $\quad=$ coding sequence ENSP00000451429
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTAWLILAVISVY,,DLVAVLCPKGPLRMLVETAQE RNETLFPALIYS,,STMV WLVNMAEGDPEAQRRVSKNSKYNAE,,RGVKLGLGDFIFYSVLVGKA SATA SGDWNTTIACFVAILI ,,GLCLTLLLLA IF KKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQFYI* 226 amino acids $=25.4 \mathrm{kDa}$
 RNETLFPALIYS,,STMVWLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ 2 $\Varangle 44^{*}$ amino acids $=31.7 \mathrm{kDa}$

```
PSEN1-232
    186 .. 26,831 26,646 bp ■ 
* 6 segments = 856 bp
/codon_start = 1
/note = coding sequence ENSP00000514904
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTAWLILAVISVY,,DLVAVLCPKGPLRMLVETAQE
        RNETLFPALIYS,,STMVWLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI
        LAGEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ
        2%4*amino acids = 31.7 kDa
PSEN1-233
    186 .. 26,831 26,646 bp \square }\quad->\quad\textrm{CDS
* 6 segments = 856 bp
/codon_start = 1
/note = coding sequence ENSP00000514905
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,DLVAVLCPKGPLRMLVETAQE
        RNETLFPALIYS,,STMV WLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI
        LAGEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ
        \Sigma&4*amino acids = 31.7 kDa
PSEN1-235 186 .. 26,831 26,646 bp \square | CDS
* 5 segments = 682 bp
/codon_start = 1
/note = coding sequence ENSP00000514906
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTAWLILAVISVY,,DLVAVLCPKGPLRMLVETAQE
        RNETLFPALIYS,,STMV WLVNMAEGDPEAQRRVSKNSKYNAE,,RGVKLGLGDFIFYSVLVGKASATA SGDWNTTIACFVAILI,,GLCLTLLLLAIF
        KKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQFYI*
        226 amino acids = 25.4 kDa
PSEN1-237
    186 .. 26,831 26,646 bp \square }->\mathrm{ CDS
* 6 segments = 856 bp
/codon_start = 1
/note = coding sequence ENSP00000514908
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,DLVAVLCPKGPLRMLVETAQE
        RNETLFPALIYS,,STMVWLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI
        LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ
        \Sigma&4*amino acids = 31.7 kDa
```

Feature
Location
Size
Type
/note $\quad=$ coding sequence ENSP00000514933
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,DLVAVLCPKGPLRMLVETAQE RNETLFPALIYS,,STMV WLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATA SGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ £ $\Varangle 44^{*}$ amino acids $=31.7 \mathrm{kDa}$

PSEN1-243
( 5 segments $=757 \mathrm{bp}$
/codon_start = 1
/note $\quad=$ coding sequence ENSP00000514934
/translation $=$ EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILA VISVY,,ATMVWLVNMAEGDPEAQRRV SKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSILAGEDPEE,,RGVKLGLGDFIFYSVLVGKASATAS GDWNTTIACFVAILI,,GLCLTLLLLA IFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQFYI* 251 amino acids $=28.0 \mathrm{kDa}$


PSEN1-253
186 .. $26,831 \quad 26,646 \mathrm{bp} \quad \square \quad \rightarrow \quad \mathrm{CDS}$

- 6 segments $=856 \mathrm{bp}$
/codon_start $=1$
/note $\quad=$ coding sequence ENSP00000514944
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,DLVAVLCPKGPLRMLVETAQE RNETLFPALIYS,,STMVWLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ £ $84^{*}$ amino acids $=31.7 \mathrm{kDa}$
PSEN1-256 186 .. 26,831 26,646 bp $\square \rightarrow$ CDS

```
* 6 segments = 883 bp
```

/codon_start $=1$
/note $\quad=$ coding sequence ENSP00000514947
/translation $=$ EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,DVFYVFLFLDLVAVLCPKGPLR MLVETAQERNETLFPALIYS,,STMVWLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRA AVQELSSSILAGEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQP



Feature
Location
Size
Type
PSEN1-258

* 6 segments $=856 \mathrm{bp}$
/codon_start = 1
/note $\quad=$ coding sequence ENSP00000514949
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,DLVAVLCPKGPLRMLVETAQE RNETLFPALIYS,,STMV WLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATA SGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ £ $\Varangle 44^{*}$ amino acids $=31.7 \mathrm{kDa}$

PSEN1-259

- 6 segments $=856 \mathrm{bp}$
/codon_start $=1$
/note $\quad=$ coding sequence ENSP00000514950
/translation $=$ EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,DLVAVLCPKGPLRMLVETAQE RNETLFPALIYS,,STMVWLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ $2 \% 4^{*}$ amino acids $=31.7 \mathrm{kDa}$
 RNETLFPALIYS,,STMV WLVNMAEGDPEAQRRVSKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSI LA GEDPEE,,RGVKLGLGDFIFYSVLVGKASATASGDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQ 2 $\Varangle 44^{*}$ amino acids $=31.7 \mathrm{kDa}$

/codon_start = 1
/note $\quad=$ coding sequence ENSP00000515001
/translation = EVFKTYNVAVDYITVALLIWNFGVVGMISIHWKGPLRLQQAYLIMISALMALVFIKYLPEWTA WLILAVISVY,,ATMVWLVNMAEGDPEAQRRV SKNSKYNAE,,STERESQDTVAENDDGGFSEEWEAQRDSHLGPHRSTPESRAAVQELSSSILAGEDPEE,,RGVKLGLGDFIFYSVLVGKASATAS GDWNTTIACFVAILI,,GLCLTLLLLAIFKKALPALPISITFGLVFYFATDYLVQPFMDQLAFHQFYI* 251 amino acids $=28.0 \mathrm{kDa}$


| Donor Template WT -> SNV | 5592 | .. | 5691 | 100 bp | $\square$ | $\mapsto$ | misc_feature |
| :--- | ---: | :--- | ---: | ---: | ---: | ---: | ---: |
| Protospacer Sequence | 5610 | .. | 5629 | 20 bp | $\square$ | $\mapsto$ | misc_feature |
| Silient SNV | 5628 | .. | 5628 | 1 bp | $\square$ | $\mapsto$ | misc_feature |


| $/$ note $\quad$ | $W T=T$ |
| ---: | :--- |
|  | SNV $=G$ |


| PAM | 5630 | .. | 5632 | 3 bp | $\square$ | $\mapsto$ | misc_feature |
| :--- | :--- | :--- | :--- | :--- | :--- | :--- | :--- |
| SNV | 5642 | .. | 5642 | 1 bp | $\square$ | $\mapsto$ | misc_feature |


/note $\quad=\quad$ primary transcript ENST00000555867 Retained intron
PSEN1-228 $\quad 24,182$.. $26,923 \quad 2742 \mathrm{bp} \quad \square \quad \rightarrow \quad$ prim_transcript
/note $\quad=\quad$ primary transcript ENST00000697915 Retained intron


