

Supplementary Figure

A

V Q L K S X
↓

agTACAACTGAAGAGTTGAGTAGTTGCTACAGAGACTGGATGGTCCACAAAGCC
TAAAGTATTTACTCTGGTCCTTTACACAAAAGGCTTACCAATACCTGGGCTAGGgt

B

D C R F T Q D T Y X
↓

agATTGCAGATTCACACAAGATACATACTGAGGGAAGTCTTAGAAGAAGAGAGGG
CAGCCTGGAGCACGAAATATTGgt

C

V V H F C L X
↓

agGTTGTACATTTTTGTTTGTGACAATTGGTCCTGCACTTTTGAGTGAAGGAAAT
AATGTTTGATAACTGCTAGAAGAATCTACCAGAAAAGATTGTGATTTCTGCTTT
CCTGTAACTTTTTGGGgt

D

G Q Q R N H Y K F X
↓

agGTCAACAGAGGAACCATTATAAGTTTTGAGCACTACGAATTCAAAAATGTAA
TGCAGTTAGAAGATGGAATCTGCTATCAGCATgtaag

Supplementary Figure. Alignment of mutation-induced cryptic exons
(A) Patient ID 19. **(B)** Patient ID 48. **(C)** Patient ID 126. **(D)** Patient ID 217.
All cryptic exons contained stop codons, meaning they are truncating mutations.