Table S3. List of primers used in the study.

|  |  |
| --- | --- |
| F108AF | 5'- CTTGCAGGGATCTGGGGCCGCACAGGTGCCAAAT -3’ |
| F108AR | 5'- ATTTGGCACCTGTGCGGCCCCAGATCCCTGCAAG -3’ |
| P109AF | 5'- CTTGCAGGGATCTGCGAACGCACAGGTGC -3’ |
| P109AR | 5'- GCACCTGTGCGTTCGCAGATCCCTGCAAG -3' |
| D110AF | 5'- ATATATTCTTGCAGGGAGCTGGGAACGCACAGGTG -3' |
| D110AR | 5'- CACCTGTGCGTTCCCAGCTCCCTGCAAGAATATAT -3' |
| P111AF | 5'- AATATATTCTTGCAGGCATCTGGGAACGCACAGGT -3' |
| P111AR | 5'- ACCTGTGCGTTCCCAGATGCCTGCAAGAATATATT -3' |
| C112AF | 5'- ACCTGGAAAATATATTCTTGGCGGGATCTGGGAACGCACAG -3' |
| C112AR | 5'- CTGTGCGTTCCCAGATCCCGCCAAGAATATATTTTCCAGGT -3' |
| K113AF | 5'- AAAAACCTGGAAAATATATTCGCGCAGGGATCTGGGAACGCAC -3' |
| K113AR | 5'- GTGCGTTCCCAGATCCCTGCGCGAATATATTTTCCAGGTTTTT -3' |
| N114AF | 5'- GAAAAAAACCTGGAAAATATAGCCTTGCAGGGATCTGGGAACGC -3' |
| N114AR | 5'- GCGTTCCCAGATCCCTGCAAGGCTATATTTTCCAGGTTTTTTTC -3' |
| I115AF | 5'- AGTAAGAAAAAAACCTGGAAAATGCATTCTTGCAGGGATCTGGGAACG -3' |
| I115AR | 5'- CGTTCCCAGATCCCTGCAAGAATGCATTTTCCAGGTTTTTTTCTTACT -3' |
| F116AF | 5'- GAAAAAAACCTGGAAGCTATATTCTTGCAGGGATCTGGGAACGC -3' |
| F116AR | 5'- GCGTTCCCAGATCCCTGCAAGAATATAGCTTCCAGGTTTTTTTC -3' |
| S117AF | 5'- GTAAGAAAAAAACCTGGCAAATATATTCTTGCAGGGATCTGGGAAC -3' |
| S117AR | 5'- GTTCCCAGATCCCTGCAAGAATATATTTGCCAGGTTTTTTTCTTAC -3' |
| R118AF | 5'- CCCTCGGAAGTAAGAAAAAAACGCGGAAAATATATTCTTGCAGGGA -3' |
| R118AR | 5'- TCCCTGCAAGAATATATTTTCCGCGTTTTTTTCTTACTTCCGAGGG -3' |
| F119AF | 5'- CACCCCTCGGAAGTAAGAAAAAGCCCTGGAAAATATATTCTTGCAG -3’ |
| F119AR | 5'- CTGCAAGAATATATTTTCCAGGGCTTTTTCTTACTTCCGAGGGGTG -3’ |
| F120AF | 5'- CCACCCCTCGGAAGTAAGAAGCAAACCTGGAAAATATATTCTTGC -3’ |
| F120AR | 5'- GCAAGAATATATTTTCCAGGTTTGCTTCTTACTTCCGAGGGGTGG -3’ |
| S121AF | 5'- CCACCCCTCGGAAGTAAGCAAAAAACCTGGAAAATATATTCTTG -3’ |
| S121AR | 5'- CAAGAATATATTTTCCAGGTTTTTTGCTTACTTCCGAGGGGTGG -3’ |
| Y122AF | 5'- ACCTCCACCCCTCGGAAGGCAGAAAAAAACCTGGAAAATATATTCT -3’ |
| Y122AR | 5'- AGAATATATTTTCCAGGTTTTTTTCTGCCTTCCGAGGGGTGGAGGT -3’ |
| F123AF | 5'-GACCTCCACCCCTCGGGCGTAAGAAAAAAACCTGGAAAATATA -3’ |
| F123AR | 5'- TATATTTTCCAGGTTTTTTTCTTACGCCCGAGGGGTGGAGGTC -3’ |
| R124AF | 5'- GTGACCTCCACCCCTGCGAAGTAAGAAAAAAACCTGGAA -3’ |
| R124AR | 5'- TTCCAGGTTTTTTTCTTACTTCGCAGGGGTGGAGGTCAC -3’ |
| G125AF | 5’- GTGACCTCCACCGCTCGGAAGTAAGAAAAAAACCTGGAA -3’ |
| G125AR | 5’- TTCCAGGTTTTTTTCTTACTTCCGAGCGGTGGAGGTCAC -3’ |
| D110VF | 5’- ATATATTCTTGCAGGGAACTGGGAACGCACAGGTG -3’ |
| D110VR | 5’- CACCTGTGCGTTCCCAGTTCCCTGCAAGAATATAT |
| P111HF | 5’- GTGCGTTCCCAGATCACTGCAAGAATATATTTTCC -3’ |
| P111HR | 5’- GGAAAATATATTCTTGCAGTGATCTGGGAACGCAC -3’ |
| N114HF | 5’- GATCCCTGCAAGCATATATTTTCCAGG -3’ |
| N114HR | 5’- CCTGGAAAATATATGCTTGCAGGGATC -3’ |
| I115TF | 5’- AAAAAAACCTGGAAAACGTATTCTTGCAGGGATCTGGGAACGC -3’ |
| I115TR | 5’- GCGTTCCCAGATCCCTGCAAGAATACGTTTTCCAGGTTTTTTTC -3’ |
| R118SF | 5’- CCCTCGGAAGTAAGAAAAAAAGCTGGAAAATATATTCTTGCAG -3’ |
| R118SR | 5’- CTGCAAGAATATATTTTCCAGCTTTTTTTCTTACTTCCGAGGG -3’ |
| S121LF | 5’- TCCAGGTTTTTTCTCTACTTCCGAGGG -3’ |
| S121LR | 5’- CCCTCGGAAGTAGAGAAAAAACCTGGA -3’ |
| 108\_125RPE65Fwd | 5’- TACTTCCAATCCAATGCATTCCCAGATCCCTGCAAG -3’ |
| 108\_125RPE65Rev | 5’- TTATCCACTTCCAATGTTATTACCCTCGGAAGTAAG-3’ |

Figure S1.

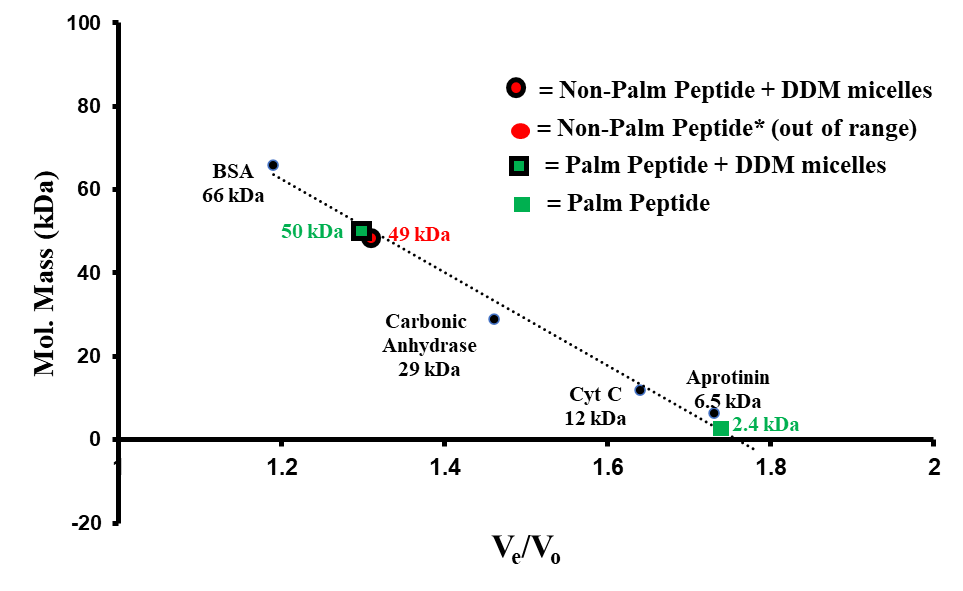
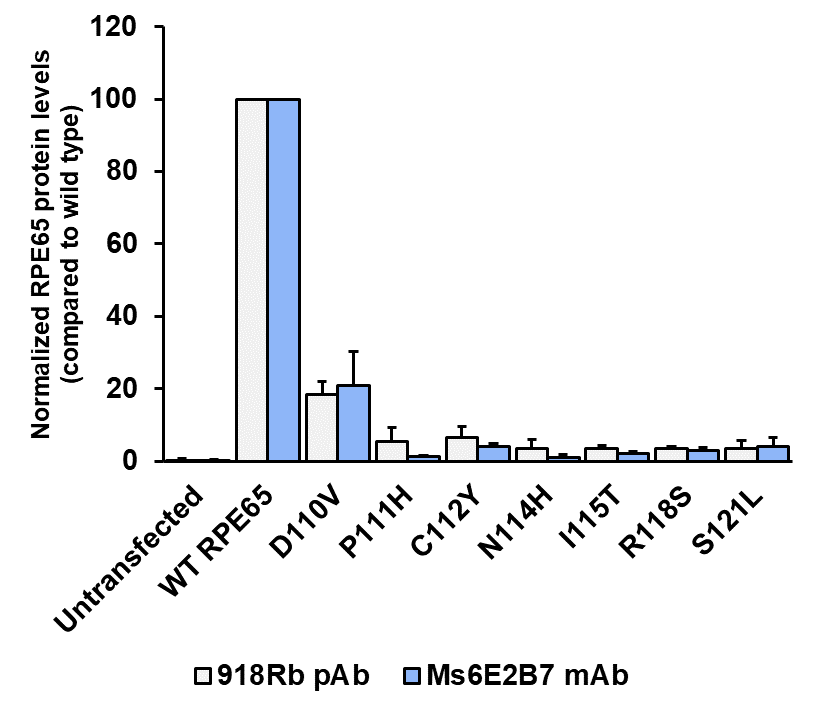
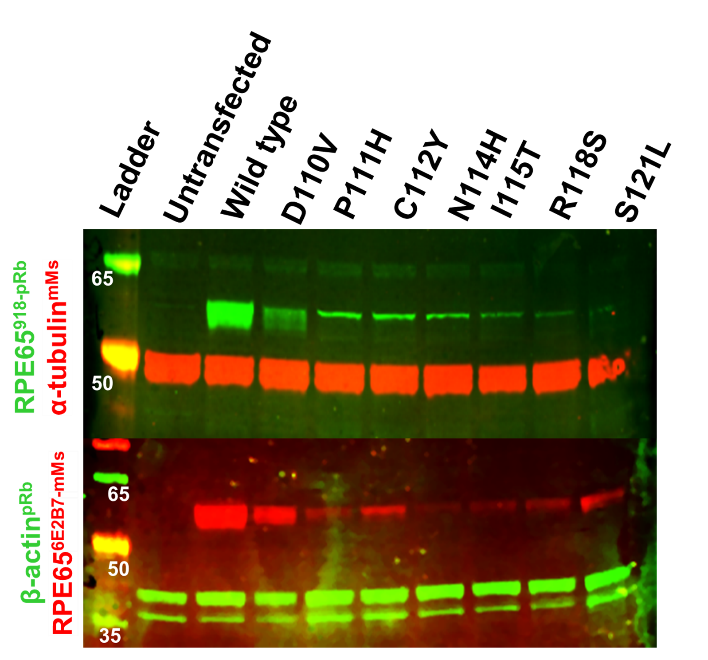


Figure S2. Pathogenic mutations/variants of unknown significance (VUS) in AH107-125 significantly impact the protein expression level of RPE65 protein.



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