

Table S1 Primer pairs for RT-qPCR analysis

Primer name	Sequences
PLXNB3-F	CGCTTCTCCGCACCTAATACC
PLXNB3-R	CAGGGCTGTCGATTACAGGG
GAPDH-F	GGAGCGAGATCCCTCCAAAAT
GAPDH-R	GGCTGTTGTCATACTTCTCATGG
Notch1-F	GAGGCGTGGCAGACTATGC
Notch1-R	CTTGTACTCCGTCAGCGTGA
Jag1-F	GTCCATGCGAAGCGTGAACG
Jag1-R	GCGGGACTGATACTCCTTGA
DLL1-F	GATTCTCCTGATGACCTCGCA
DLL1-R	TCCGTAGTAGTGTTCGTCACA
HES1-F	TCAACACGACACCGGATAAAC
HES1-R	GCCGCGAGCTATCTTTCTTCA
HEY1-F	GTTCCGGCTCTAGGTTCCATGT
HEY1-R	CGTCGGCGCTTCTCAATTATTC
UQCRB-F	GGTAAGCAGGCCGTTTCAG
UQCRB-R	AGGTCCAGTGCCCTCTTAATG
UQCRQ-F	CGCGAGTTTGGGAATCTGAC
UQCRQ-R	TAGTGAAGACGTGCGGATAGG
COX7A2-F	CTCGGAGGTAGTTCGGTTC
COX7A2-R	TCTGCCCAATCTGACGAAGAG
UQCR10-F	ATCGTGGGCGTCATGTTCTTC
UQCR10-R	ATGTGGTCGTAGATAGCGTCC
COX6B1-F	CTACAAGACCGCCCTTTTGA
COX6B1-R	GCAGAGGGACTGGTACACAC
COX5B-F	ATGGCTTCAAGGTTACTTCGC
COX5B-R	CCCTTTGGGGCCAGTACATT
ATP1B3-F	AACCCGACCACCGGAGAAT
ATP1B3-R	TGAGAGTCTGAAGCATAACCCA
CACNG5-F	GGCGTTGCTTCACCATAGAAT
CACNG5-R	GCTGAGCGGATCATCTTTAGAAC
CREBBP-F	CAACCCCAAAGAGCCAAACT
CREBBP-R	CCTCGTAGAAGCTCCGACAGT
ATP5E-F	GGCTGGACTCAGCTACATCC
ATP5E-R	TTACGTTGCTGCCAGAAGTCT
EP300-F	AGCCAAGCGGCCTAAACTC
EP300-R	TCACCACCATTGGTTAGTCCC
HTT-F	AAACTTCTGGGCATCGCTATG
HTT-R	GTTGAGGCATTCGTCAGCCA
UQCRH-F	GAGGACGAGCAAAAGATGCTT
UQCRH-R	CGAGAGGAATCACGCTCATCA
LRP1-F	CTATCGACGCCCTAAGACTT
LRP1-R	CATCGCTGGGCCTTACTCT
NDUFS3-F	ACTGTCAGACCACGGAATGAT
NDUFS3-R	GGGCAAGATTTAGCCACATAC

RT-qPCR, reverse transcription-quantitative polymerase chain reaction.

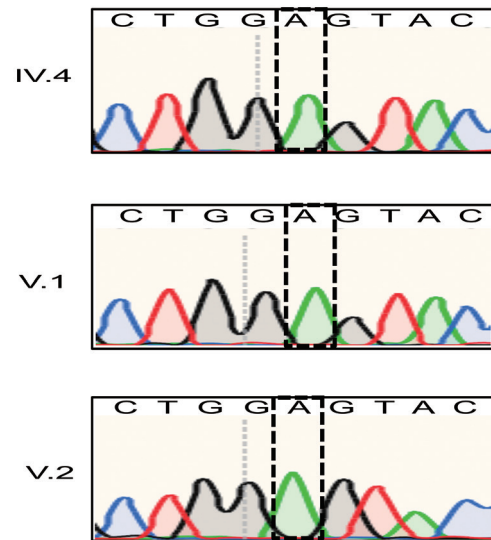


Figure S1 Sanger sequencing in the other members of the Yunnan family for confirmation of the c.A4319T mutation.

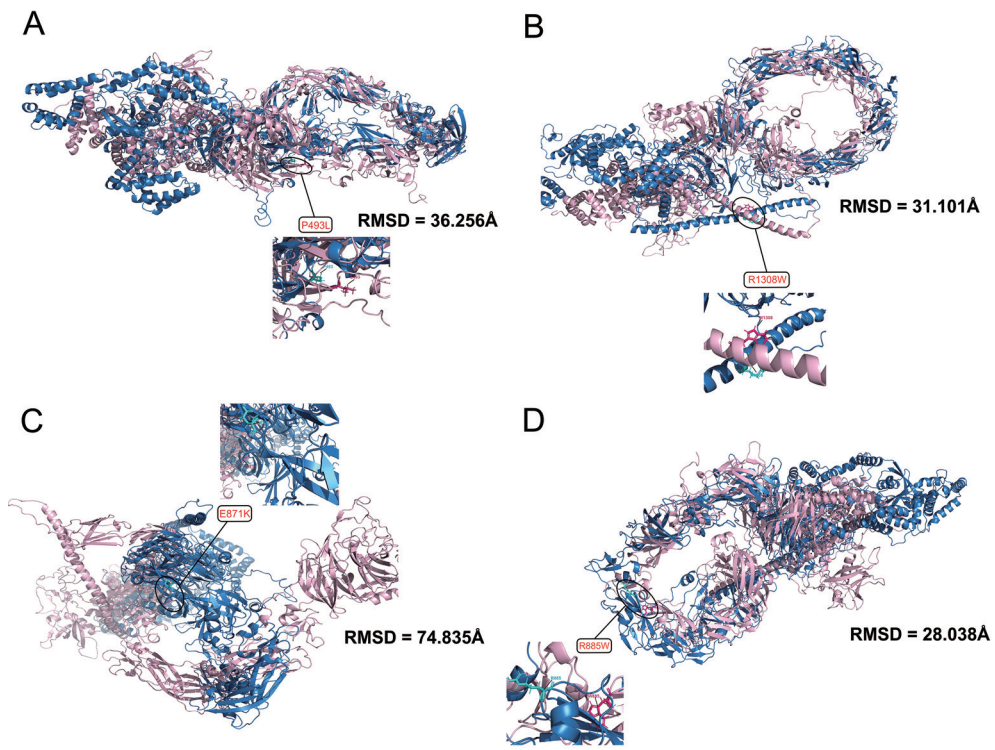


Figure S2 Structural changes in PLXNB3 protein caused by four mutations predicted by AlphaFold and PyRosetta.