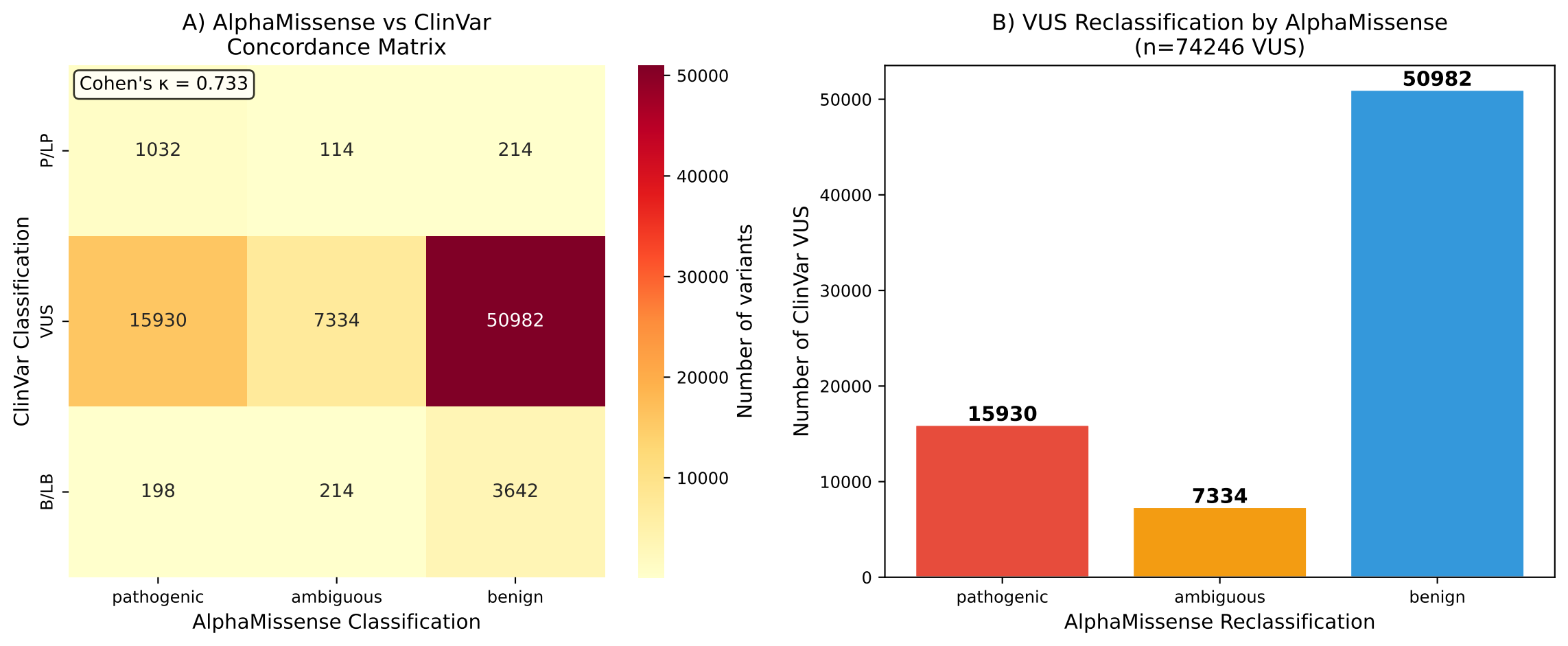
A) AlphaMissense vs ClinVar
Concordance Matrix
Cohen's κ = 0.733
1032
114
214
15930
7334
50982
198
214
3642
P/LP
VUS
B/LB
ClinVar Classification
pathogenic
ambiguous
benign
AlphaMissense Classification
50000
40000
30000
20000
10000
Number of variants
B) VUS Reclassification by AlphaMissense
(n=74246 VUS)
Number of ClinVar VUS
0
10000
20000
30000
40000
50000
15930
7334
50982
pathogenic
ambiguous
benign
AlphaMissense Reclassification