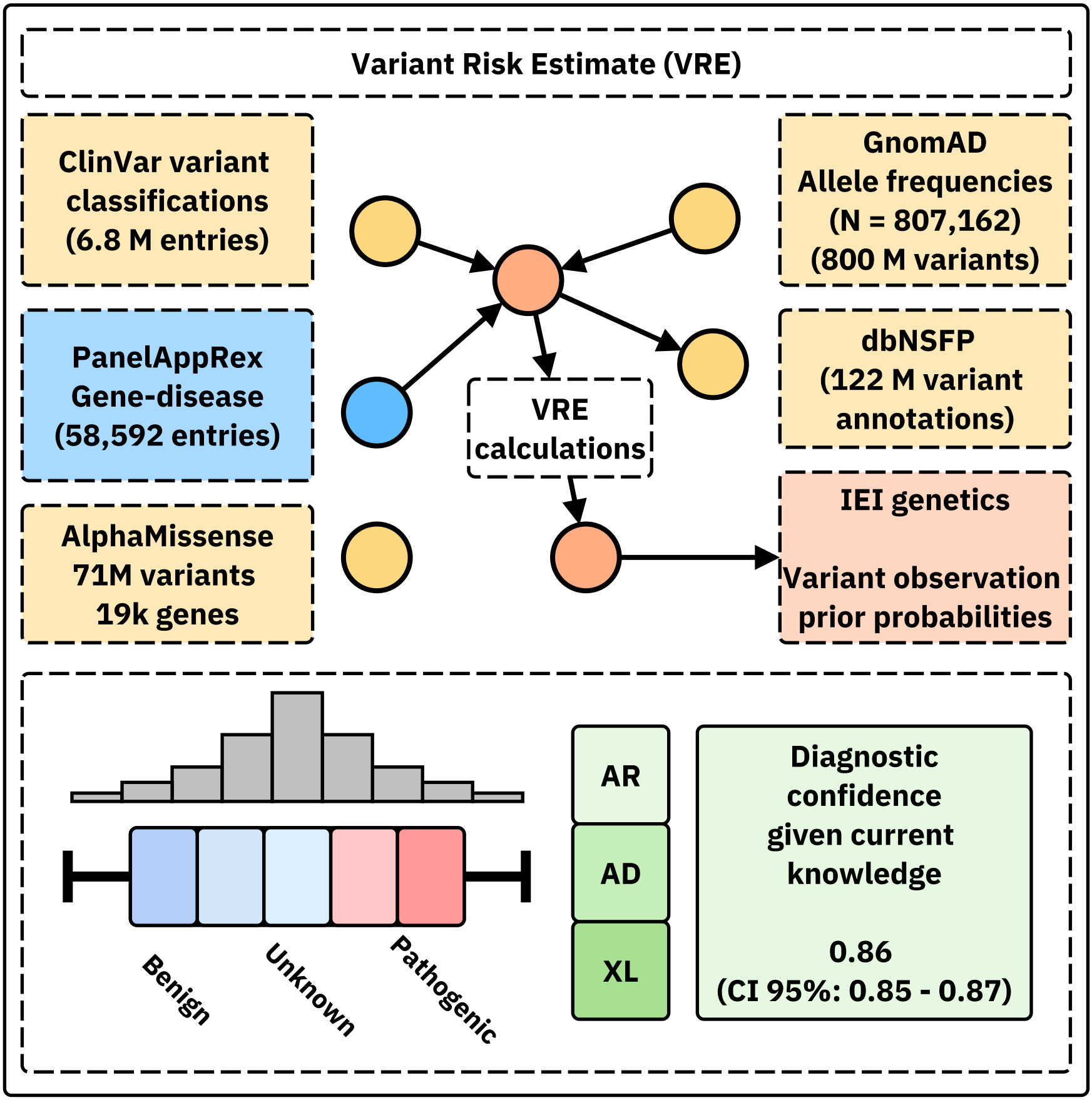
Variant Risk Estimate (VRE)
ClinVar variant classifications (6.8 M entries)
PanelAppRex Gene-disease (58,592 entries)
AlphaMissense 71M variants 19k genes
VRE calculations
GnomAD Allele frequencies (N = 807,162) (800 M variants)
dbNSFP (122 M variant annotations)
IEI genetics
Variant observation prior probabilities
Benign
Unknown
Pathogenic
AR
AD
XL
Diagnostic confidence given current knowledge
0.86
(CI 95%: 0.85 - 0.87)