



Figure S4. Phenoxome ROC Curves of Different α Values Based on Pathogenic Variant Ranks

The ROC curves were generated using clinical samples in the study. Sensitivity was defined as the frequency of pathogenic variants that were ranked above a particular threshold position, and specificity as the percentage of pathogenic variants ranked below the threshold. The AUC scores for $\alpha=0.7$, 0.8 and 0.9 are 0.981, 0.984 and 0.982 respectively.