

Table S6. Performance of prediction tools with PMD testing dataset.

	MAPP	nsSNPAnalyzer	PANTHER	PhD-SNP	PPH-1	PPH-2	SIFT	SNAP	PredictSNP
True positives	1,353	892	390	1,521	1,468	1,846	1,379	1,345	1,738
False negatives	294	589	262	728	709	357	275	781	511
True negatives	576	482	478	735	773	526	480	756	688
False positives	613	253	202	513	445	710	579	444	560
Total	2,836	2,216	1,332	3,497	3,395	3,439	2,713	3,326	3,497
Sensitivity ^a	0.821	0.602	0.598	0.676	0.674	0.838	0.834	0.633	0.773
Specificity ^a	0.484	0.656	0.703	0.589	0.635	0.426	0.453	0.630	0.551
Precision ^a	0.614	0.636	0.668	0.622	0.649	0.593	0.604	0.631	0.633
NPV ^a	0.731	0.622	0.636	0.645	0.661	0.724	0.732	0.632	0.708
Accuracy ^a	0.653	0.629	0.651	0.633	0.654	0.632	0.643	0.631	0.662
MCC ^a	0.327	0.258	0.303	0.266	0.309	0.288	0.310	0.263	0.332
AUC ^a	0.695	0.630	0.697	0.676	0.658	0.704	0.685	0.667	0.720

PPH-1 – PolyPhen-1; PPH-2 – PolyPhen-2; NPV – negative predictive value; MCC – Matthews correlation coefficient; AUC – area under receiver operating characteristics curve; ^a – these metrics were calculated with normalized numbers