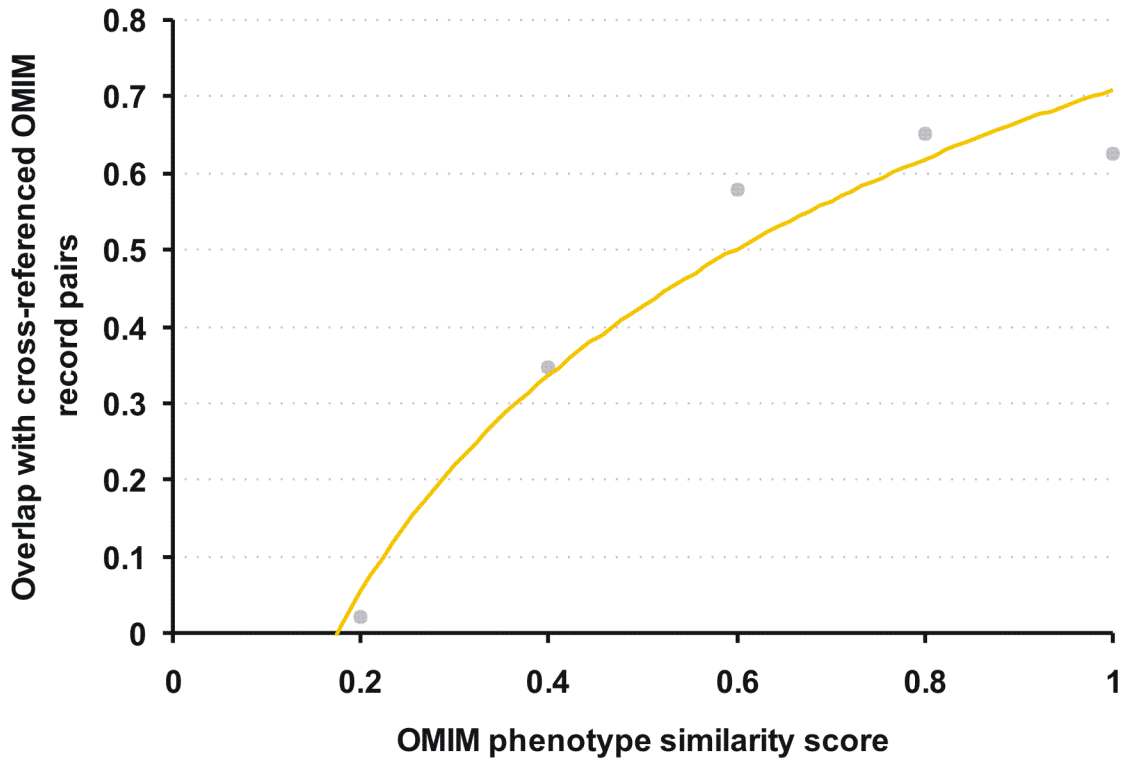


Correlation OMIM phenotype association scores and cross-referenced OMIM record pairs



Supplementary Figure 2 Benchmark of the phenotype association score. We fitted a calibration curve of the phenotype association score against the overlap with cross-referenced OMIM record pairs, that have a high degree of phenotypic overlap. Our phenotype similarity score directly correlates with the probability of overlap with these file pairs, showing that the score is a direct and reliable measure of phenotypic overlap between the records represented by the vectors.