

Supplementary Figures

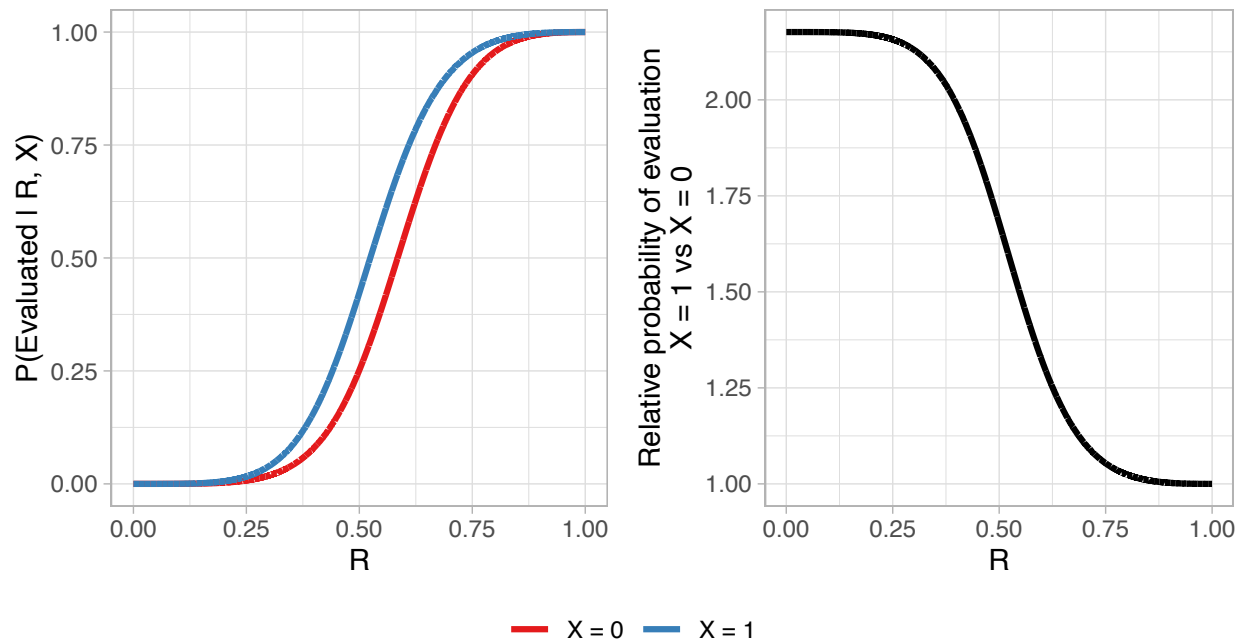


Figure S1: Visualization of clinical selection process model for Case 2B. (Right) Probability of evaluation given R and X for Case 2B ('true risk factor'). Note that while the form of the curve may appear logistic, it is not (see supplementary additional detail for clinical selection process, Cases 1-3). (Left) Relative probability of evaluation in Case 2B model for individuals with $X = 1$ vs $X = 0$ for different levels of R . When R is low (symptoms are not very representative of the disease), people with $X = 1$ are more than twice as likely to have the disease (and thus more than twice as likely to be evaluated) than people with $X = 0$. As R increases, the relative rates of evaluation converge. When R is close to 1 (symptoms are highly representative of the disease), the likelihood of disease is close to 1 in both groups, so the relative difference in likelihood of disease converges to 0. That is, for more representative cases, X is less informative to likelihood of disease.

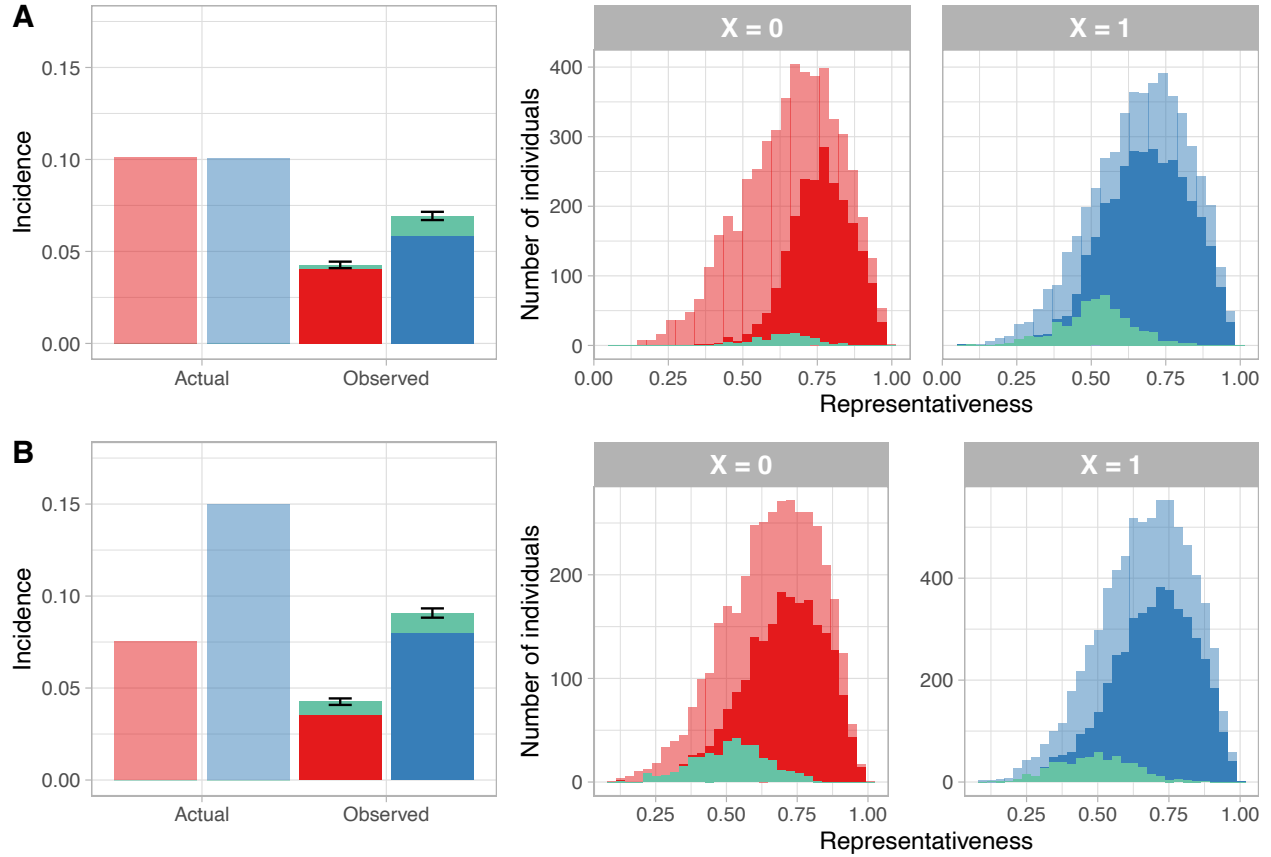


Figure S2: Ground truth disease characteristics compared with a naive analysis in Cases 2A and 2B (‘true risk factor’ and ‘false risk factor’, respectively) for a simulation in which the diagnostic evaluation has a sensitivity and specificity of 0.7. Observed incorrect diagnoses of non-diseased individuals with a false-positive evaluation result are shaded in green. In the ‘true risk factor’ case (A), individuals in the $X = 1$ group are evaluated disproportionately more often. This means that a larger share of false positive diagnoses are present for the observed data in this group. The false positive diagnoses tend to have less representative symptoms than the true positive diagnoses, distorting the distribution of observed cases. In this case, this distortion is more pronounced in the more-evaluated $X = 1$ group. (B) In the ‘false risk factor’ case, the $X = 0$ group also includes a larger number of false positives, and false positives in both groups tend to fall on the lower end of the distribution of representativeness among diagnosed cases. However, the probability of evaluation corresponds to true disease risk based on representativeness and X .