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Metric	Formula	Description
Sensitivity (Recall)	$\frac{TP}{TP+FN}$	Correctly predicted pathogenic variants divided by total pathogenic cases.
Specificity	$\frac{TN}{TN+FP}$	Correctly predicted benign variants divided by total benign cases.
PPV (Precision)	$\frac{TP}{TP+FP}$	True pathogenic predictions among all pathogenic predictions. Indicates how likely a pathogenic prediction is a TP.
NPV	$\frac{TN}{TN+FN}$	True benign predictions among all benign predictions. Indicates how likely a benign prediction is a TN.
Accuracy	$\frac{TP+TN}{TP+FP+FN+TN}$	Ratio of correct predictions to total predictions made.
MCC	$\frac{TP \cdot TN - FP \cdot FN}{\sqrt{(TP+FN)(TN+FP)(TP+FP)(TN+FN)}}$	Balanced measure of correlation between predictions and true cases. Ranges from -1 (total disagreement) to 1 (perfect agreement), with 0 indicating random predictions.