

Table 4: Confusion matrix and statistical measures of the ABCA4 DI variants.

Tool	Missing values	TP	FP	TN	FN	Accuracy	PPV	Sensitivity	Specificity	NPV	MCC
Alamut Consensus 3/4	0	11	10	50	10	75%	52%	52%	83%	83%	0.36
CADD	0	12	24	36	9	59%	33%	57%	60%	80%	0.15
DSSP	0	12	23	37	9	60%	34%	57%	62%	80%	0.17
GeneSplicer	0	13	21	39	8	64%	38%	62%	65%	83%	0.24
MaxEntScan	0	15	15	45	6	74%	50%	71%	75%	88%	0.42
NNSplice	0	13	13	47	8	74%	50%	62%	78%	85%	0.38
SpliceAI	0	19	3	57	2	94%	86%	90%	95%	97%	0.84
SpliceRover	0	15	16	44	6	73%	48%	71%	73%	88%	0.40
SpliceSiteFinder-like	0	11	27	33	10	54%	29%	52%	55%	77%	0.06

FN – false negatives, FP – false positives, MCC – Mathew’s correlation coefficient, NPV – negative predictive value - PPV – positive predictive value, TN – true negatives, TP – true positives