

Table 3: Confusion matrix and statistical measures of the *ABCA4* NCSS variants

| Tool | Missing values | TP | FP | TN | FN | Accuracy | PPV | Sensitivity | Specificity | NPV | MCC |
|---------------------------------|----------------|----|----|----|----|----------|-----|-------------|-------------|-----|-------|
| Alamut Consensus 3/4 | 0 | 43 | 2 | 5 | 21 | 68% | 96% | 67% | 71% | 19% | 0.24 |
| CADD | 0 | 17 | 3 | 4 | 47 | 30% | 85% | 27% | 57% | 8% | -0.11 |
| DSSP | 0 | 51 | 2 | 5 | 13 | 79% | 96% | 80% | 71% | 28% | 0.35 |
| GeneSplicer | 0 | 39 | 2 | 5 | 25 | 62% | 95% | 61% | 71% | 17% | 0.20 |
| MaxEntScan | 0 | 47 | 2 | 5 | 17 | 73% | 96% | 73% | 71% | 23% | 0.29 |
| MMSplice | 5 | 50 | 3 | 4 | 14 | 76% | 94% | 78% | 57% | 22% | 0.24 |
| MTSplice | 0 | 45 | 2 | 5 | 19 | 70% | 96% | 70% | 71% | 21% | 0.26 |
| NNSplice | 0 | 46 | 2 | 5 | 18 | 72% | 96% | 72% | 71% | 22% | 0.28 |
| SCAP | 28 | 40 | 2 | 5 | 24 | 63% | 95% | 63% | 71% | 17% | 0.21 |
| Spidex | 5 | 20 | 2 | 5 | 44 | 35% | 91% | 31% | 71% | 10% | 0.02 |
| SpliceAI | 0 | 50 | 1 | 6 | 14 | 79% | 98% | 78% | 86% | 30% | 0.42 |
| SpliceRover | 0 | 46 | 2 | 5 | 18 | 72% | 96% | 72% | 71% | 22% | 0.28 |
| SpliceSiteFinder-like | 0 | 44 | 2 | 5 | 20 | 69% | 96% | 69% | 71% | 20% | 0.25 |

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