**S1 Table: Pathogenicity prediction of three missense variants using three computational tools**

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| --- | --- | --- | --- |
| Variants | Mutation taster | PROVEAN | SIFT |
| Score | Prediction | Score | Prediction | Score | Prediction |
| p.Val215Gly | 4.57 | Disease causing | -7.00 | Deleterious | 0.000 | Damaging |
| p.Arg282Gln | 5.23 | Disease causing | -4.00 | Deleterious | 0.000 | Damaging |
| p.Val321Gly | 4.593 | Disease causing | -6.95 | Deleterious | 0.000 | Damaging |