**Table e-2: Results of multiple *in silico* predicting algorithms for missense variants in 33 patients with a confirmed molecular diagnosis**.

Legend: [SIFT](http://sift.bii.a-star.edu.sg/#_blank) (D = damaging, T = tolerated) (https://sift.bii.a-star.edu.sg/); [PolyPhen-2](http://genetics.bwh.harvard.edu/pph2/#_blank) (D = probably damaging, P = possibly damaging, B = benign) (<http://genetics.bwh.harvard.edu/pph2/>); MutationTaster (A = disease causing automatic, D = disease causing, N = polymorphism, P = polymorphism automatic) (<http://www.mutationtaster.org/>); FATHMM (D = damaging, T = tolerated) (http://fathmm.biocompute.org.uk/); [Likelihood ratio test (LRT)](http://www.genetics.wustl.edu/jflab/lrt_query.html#_blank) (D = deleterious, N= Neutral, U= unknown) (<http://www.genetics.wustl.edu/jflab/lrt_query.html>); MutationAssessor (L=low, M=medium, H=high) (<https://varsome.com/>); VEST3 (<http://hg19.cravat.us/CRAVAT/>); CADD\_phred (>15 Pathogenic) (https://cadd.gs.washington.edu/); GERP++ (It ranges from -12.3 to 6.17, with 6.17 being the most conserved) (<https://varsome.com/>); UMD-Predictor (<http://umd-predictor.eu/>)

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **ID** | **Gene** | **Mutations**  **cDNA level** | **Mutations**  **Protein level** | **SIFT** | **Polyphen2** | **Mutation**  **Taster** | **FATHMM** | **LRT** | **Mutation**  **Assessor** | **VEST3** | **CADD** | **GERP++** | **UMD predictor** |
| P1 | *RYR1* | c.8888T>C | p.Leu2963Pro | D | B | D | T | U | M | 0.86 | 28.7 | 4.15 | Pathogenic |
| P2 | *RYR1* | c.7373G>A | p.Arg2458His | D | P | D | D | U | M | 0.92 | 29.8 | 3.99 | Pathogenic |
| P3 | *RYR1* | c.1163C>T | p.Ser388Leu | D | P | N | D | D | L | 0.567 | 9.333 | 4.07 | Pathogenic |
| P4 | *GMPPB* | c.95C>T | p.Pro32Leu | D | D | D | D | D | H | 0.904 | 26.0 | 3.95 | Pathogenic |
| P4 | *GMPPB* | c.727C>T | p.Arg243Trp | D | B | D | T | T | L | 0.286 | 7,001 | 1.79 | Probably pathogenic |
| P5 | *MYH7* | c.2009T>C | p.Val670Ala | D | D | D | D | D | H | 0.872 | 27.2 | 4.89 | Pathogenic |
| P6 | *RYR1* | c.7291G>A | p.Asp2431Asn | D | D | D | D | D | M | 0.41 | 15.92 | 3.99 | Pathogenic |
| P7 | *RYR1* | c.5036G>A | p.Arg1679His | D | D | D | D | D | M | 0.902 | 28.7 | 4.07 | Polymorphism |
| P8 | *RYR1* | c.7048G>A | p.Ala2350Thr | D | D | N | D | D | M | 0.871 | 25.5 | 3.82 | Pathogenic |
| P9 | *ANO5* | c.1733T>C | p.Phe578Ser | D | P | D | T | N | H | 0.982 | 33 | 6.0399 | Pathogenic |
| P10 | *CAPN3* | c.633G>T | p.Lys211Asn | D | P | D | D | U | H | 0.755 | 34 | 5.4099 | Pathogenic |
| P11 | *DYSF* | c.862G>T | p.Asp288Tyr | D | D | D | D | D | M | 0.962 | 26.3 | 4.5999 | - |
| P11 | *DYSF* | c.2875C>T | p.Arg959Trp | D | D | D | D | D | L | 0.947 | 33 | 5.21 | - |
| P12 | *RYR1* | c.6599C>T | p.Ala2200Val | T | P | D | D | T | L | 0.896 | 21 | 4.59 | Pathogenic |
| P14 | *ANO5* | c.580C>T | p.Arg194Trp | D | D | D | T | T | M | 0.685 | 32 | 4.7699 | Probably pathogenic |
| P14 | *ANO5* | c.2219C>T | p.Ser740Phe | D | D | D | T | D | H | 0.987 | 33 | 5.25 | - |
| P17 | *CPT2* | c.338C>T | p.Ser113Leu | D | D | D | D | D | M | 0.935 | 34 | 5.88 | Probably pathogenic |
| P18 | *RYR1* | c.4711A>G | p.Ile1571Val | T | D | N | D | D | N | 0.718 | 3.864 | 3.99 | Polymorphism |
| P18 | *RYR1* | 10097G>A | p.Arg3366His | D | P | D | D | D | L | 0.686 | 23.9 | 3.54 | Polymorphism |
| P18 | *RYR1* | c.11798A>G | p.Tyr3933Cys | D | D | D | D | D | M | 0.951 | 24.1 | 4.11 | Probably pathogenic |
| P19 | *CAPN3* | c.2257G>A | p.Asp753Asn | T | D | D | D | D | L | 0.394 | 24.6 | 4.5399 | Pathogenic |
| P19 | *CAPN3* | 1453A>G | p.Met485Val | D | P | D | D | D | M | 0.935 | 24.9 | 4.5399 | Pathogenic |
| P20 | *SGCA* | c.242G>A | p.Arg81His | D | D | D | D | D | M | 0.546 | 28.8 | 4.53 | Probably pathogenic |
| P20 | *SGCA* | c.739G>A | p.Val247Met | D | D | D | D | D | L | 0.738 | 17.6 | 3.85 | Probable polymorphism |
| P23 | *RYR1* | c.14812A>G | p.Ile4938Val | D | P | D | D | D | M | 0.669 | 9.658 | 4.67 | Pathogenic |
| P24 | *MYOT* | c.179C>T | p.Ser60Phe | D | P | D | T | T | M | 0.525 | 28.8 | 6.02 | Pathogenic |
| P26 | *POMT2* | c.1733G>A | p.Arg578His | T | B | D | D | D | M | 0.718 | 23.2 | 4.79 | Pathogenic |
| P26 | *POMT2* | c.707T>G | p.Leu236Arg | D | D | D | D | D | H | 0.998 | 29.2 | 5.75 | Pathogenic |
| P26 | *POMT2* | 239C>T | p.Pro80Leu | T | B | D | D | T | M | 0.27 | 15.82 | -1.87 | Probably pathogenic |
| P27 | *PYGM* | c.406G>A | p.Gly136Ser | D | D | D | D | D | H | 0.976 | 27.1 | 5.47 | Pathogenic |
| P28 | *SGCA* | c.739G>A | p.Val247Met | D | D | D | D | D | M | 0.738 | 25.8 | 3.8499 | Probable polymorphism |
| P28 | *SGCA* | c.850C>T | p.Arg284Cys | D | D | D | D | D | M | 0.931 | 33 | 5.8 | Polymorphism |
| P30 | *ANO5* | c.629C>T | p.Ser210Leu | D | D | D | T | D | M | 0.864 | 34 | 5.69 | - |
| P31 | *RYR1* | c.5288C>T | p.Pro1763Leu | D | D | D | T | D | M | 0.439 | 27.8 | 3.77 | Pathogenic |
| P31 | *RYR1* | c.7681C>T | p.Leu2561Phe | D | D | D | D | D | M | 0.423 | 23.3 | 1.81 | Probably pathogenic |