**Table e-3. Reference allele frequencies in polymorphic database** (gnomAD, http://gnomad.broadinstitute.org) **for missense variants identified in molecularly defined patients.**\* calculated maximum tolerated allele counts (using http://cardiodb.org/alleleFrequencyApp)

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **ID** | **Gene** | **Mutations**  **cDNA Protein** | | **Allele count** | **Allele frequency** | **Maximum tolerated reference allele count in selected neuromuscular genes\*** |
| P1 | *RYR1* | c.8888T>C | p.Leu2963Pro | 2 | 0.000007953 | 241 |
| P2 | *RYR1* | c.7373G>A | p.Arg2458His | 2 | 0.000007961 |  |
| P3 | *RYR1* | c.1163C>T | p.Ser388Leu | 6 | 0.00002387 |  |
| P4 | *GMPPB* | c.95C>T | p.Pro32Leu | 18 | 0.00008002 | 32 |
| P4 | *GMPPB* | c.727C>T | p.Arg243Trp | 7 | 0.00002787 |  |
| P5 | *MYH7* | c.2009T>C | p.Val670Ala | 0 | 0 | 18 |
| P6 | *RYR1* | c.7291G>A | p.Asp2431Asn | 2 | 0.000007959 |  |
| P7 | *RYR1* | c.5036G>A | p.Arg1679His | 332 | 0.001198 |  |
| P8 | *RYR1* | c.7048G>A | p.Ala2350Thr | 0 | 0 |  |
| P9 | *ANO5* | c.1733T>C | p.Phe578Ser | 54 | 0.0001911 | 39 |
| P10 | *CAPN3* | c.633G>T | p.Lys211Asn | 1 | 0.000003977 | 65 |
| P11 | *DYSF* | c.862G>T | p.Asp288Tyr | 0 | 0 | 15 |
| P11 | *DYSF* | c.2875C>T | p.Arg959Trp | 0 | 0 |  |
| P12 | *RYR1* | c.6599C>T | p.Ala2200Val | 11 |  |  |
| P14 | *ANO5* | c.580C>T | p.Arg194Trp | 0 | 0 |  |
| P14 | *ANO5* | c.2219C>T | p.Ser740Phe | 0 | 0 |  |
| P17 | *CPT2* | c.338C>T | p.Ser113Leu | 393 | 0.001390 | 428 |
| P18 | *RYR1* | c.4711A>G | p.Ile1571Val | 170 | 0.0008548 |  |
| P18 | *RYR1* | c.10097G>A | p.Arg3366His | 243 | 0.0008605 |  |
| P18 | *RYR1* | c.11798A>G | p.Tyr3933Cys | 240 | 0.0008487 |  |
| P19 | *CAPN3* | c.2257G>A | p.Asp753Asn | 210 | 0.0007442 |  |
| P19 | *CAPN3* | c.1453A>G | p.Met485Val | 0 | 0 |  |
| P20 | *SGCA* | c.242G>A | p.Arg81His | 1 | 0.000004002 | 131 |
| P20 | *SGCA* | c.739G>A | p.Val247Met | 35 | 0.0001242 |  |
| P23 | *RYR1* | c.14812A>G | p.Ile4938Val | 0 | 0 |  |
| P24 | *MYOT* | c.179C>T | p.Ser60Phe | 10 | 0.00003566 | 11 |
| P26 | *POMT2* | c.1733G>A | p.Arg578His | 12 | 0.00004826 | 23 |
| P26 | *POMT2* | c.707T>G | p.Leu236Arg | 0 | 0 |  |
| P26 | *POMT2* | c.239C>T | p.Pro80Leu | 0 | 0 |  |
| P27 | *PYGM* | c.406G>A | p.Gly136Ser | 0 | 0 |  |
| P28 | *SGCA* | c.739G>A | p.Val247Met | 35 | 0.0001242 |  |
| P28 | *SGCA* | c.850C>T | p.Arg284Cys | 42 | 0.0001485 |  |
| P30 | *ANO5* | c.629C>T | p.Ser210Leu | 0 | 0 |  |
| P31 | *RYR1* | c.5288C>T | p.Pro1763Leu | 0 | 0 |  |
| P31 | *RYR1* | c.7681C>T | p.Leu2561Phe | 2 | 0.000008000 |  |