**Supplementary tables:**

**Table-1:** Novel variants and their ACMG prediction:

|  |  |
| --- | --- |
| **Variant** | **Prediction** |
| c.445G>A, p.Ala149Thr | LP |
| c.925\_927delinsTTGGGAT, p.Ala309LeufsTer4 | P |
| c.1474\_1475insCAGCAGAATCTG, p.Ala491\_Val492insAlaAlaGluSer | LP |
| c.1783G>A, p.Ala595Thr | LP |
| c.2005G>A, p.Ala669Thr | LP |
| c.533G>A, p.Arg178Lys | LP |
| c.384T>G\*, p.Asp128Glu | LP |
| c.729\_738delAGATTACATT, p.Asp244LeufsTer7 | P |
| c.1089T>A, p.Asp363Glu | P |
| c.1451G>A, p.Cys484Tyr | LP |
| c.1111C>T, p.Gln371Ter | P |
| c.1153C>T, p.Gln385Ter | P |
| c.425G>T, p.Gly142Val | LP |
| c.1685G>A, p.Gly562Glu | LP |
| c.1837G>A, p.Gly613Arg | LP |
| c.752A>C, p.His251Pro | LP |
| c.1748A>G, p.His583Arg | LP, P, LP |
| c.477C>G, p.Ile159Met | LP |
| c.516\_519 dupCATT, p.Ile173HisfsTer3 | P |
| c.1760T>C, p.Ile587Thr | P |
| c.2058\_2059del, p.Ile687ProfsTer47 | P |
| c.1919T>G, p.Leu640Arg | LP |
| c.215T>G, p.Leu72Arg | LP |
| c.214C>T\*, p.Leu72Phe | LP |
| c.2196G>C, p.Leu732Phe | LP |
| c.657A>C, p.Lys219Asn | LP |
| c.723del, p.Lys241AsnfsTer13 | P |
| c.18989delA, p.Lys633SerfsTer41 | LP |
| c.1778\_1780del, p.Phe593del | LP |
| c.816\_820del, p.Ser273Ter | P |
| c.1313dup, p.Ser439LysfsTer6 | P |
| c.1368C>G\*, p.Ser456Arg | LP |
| c.544A>G, p.Thr182Ala | LP |
| c.575C>T, p.Thr192Ile | LP |
| c.1709C>A\*, p.Thr570Lys | LP |
| c.263C>T, p.Thr88Ile | LP |
| c.1061\_1064dup, p.Tyr355Ter | P |
| c.1154\_1157dup, p.Tyr386Ter | P |
| c.1006del, p.Val336PhefsTer11 | P |
| c.2051T>A , p.Val684Asp | LP |
| c.52-3C>G, 3 splice site (intron1) | VUS |
| c.52-1G>T (Intron-1) | P |
| c.1075+2T>C (Intron-5) | P |
| c.1374+1\_1374+2del (Intron-7) | P |
| c.1909+2T>C (Intron-10) | P |
| c.1727-2A>G (Intron-9) | P |

***Footnotes:*** *P-Pathogenic; LP-Likely pathogenic*

**Supplementary table – 2:** Comparison with previous studies

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Study author/year** | **No. of patients included in study** | **Age at onset (Mean ± SD)** | **Age at diagnosis (Mean ± SD)** | **No. of wheel chair bound patients**  **No (%)** | **Mean age at wheelchair dependence/Mean duration** |
| Mori- Yoshimura et al /2014 [17] | 121 | 27.9 ± 9.6 | 44.9 ± 13.9 | 78 (64) | -/15.2 ± 0.8 |
| Pogoryelova O et al/2018 [6] | 269 | 27.8 | 32.7 | 98 (36.4) | 38.3 / 11.9 |
| Lochmuller H et al /2021 [20] | 269 | 26.8 ± 8.5 | 33.7 ± 10.8 | 39 (39) | - |
| Current study | 157 | 26.5 ± 6.2 | 32.8 ± 7.8 | 14 (8.9) | 32.0 ± 7.1 / 6.3 ± 4.9 |

**Supplementary Table -3:** Data on genotype characteristics of the cohort:

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient ID** | **variant1** | **Protein change – allele -1** | **Variant - 2** | **Protein change – allele -2** | **Domains** | **Reported/novel Variant - 1** | **Reported/novel Variant - 2** | **Pathogenicity Variant – 1 (ACMG)** | **Pathogenicity Variant – 2 (ACMG)** | **Zygosity** |
| P1 | c.2179G>A | p.Val727Met | c.1154\_1157dup | p.Tyr386Ter | K+E | Reported | Novel | LP | P | CH |
| P2 | c.2179G>A | p.Val727Met | c.2051T>A | p.Val684Asp | K+K | Reported | Novel | LP | LP | CH |
| P3 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P4 | c.2179G>A | p.Val727Met | c.1174T>C | p.Tyr392His | K+E | Reported | Reported | LP | LP | CH |
| P5 | c.2196G>C | p.Leu732Phe | c.2196G>C | p.Leu732Phe | K+K | Novel | Novel | LP | LP | HM |
| P6 | c.196G>A | p.Glu66Lys | c.196G>A | p.Glu66Lys | E+E | Reported | Reported | LP | LP | HM |
| P7 | c.2179G>A | p.Val727Met | c.1863C>A | p.Tyr621Ter | K+K | Reported | Reported | LP | P | CH |
| P8 | c.2179G>A | p.Val727Met | c.829C>T | p.Arg277Trp | K+E | Reported | Reported | LP | P | CH |
| P9 | c.2179G>A | p.Val727Met | c.1010G>A | p.Arg337Gln | K+E | Reported | Reported | LP | LP | CH |
| P10 | c.2179G>A | p.Val727Met | c.173C>T | p.Pro58Leu | K+E | Reported | Reported | LP | LP | CH |
| P11 | c.1981G>A | p.Ala661Thr | c.1981G>A | p.Ala661Thr | K+K | Reported | Reported | LP | LP | HM |
| P12 | c.2179G>A | p.Val727Met | c.2196G>C\* | p.Leu732Phe | K+K | Reported | Novel | LP | LP | CH |
| P13 | c.2179G>A | p.Val727Met | c.816\_820del | p.Ser273Ter | K+E | Reported | Novel | LP | P | CH |
| P14 | c.577C>T | p.Arg193Cys | c.577C>T | p.Arg193Cys | K+K | Reported | Reported | LP | LP | HM |
| P15 | c.2179G>A | p.Val727Met | c.52-1G>T | - | K+E | Reported | Novel | LP | P | CH |
| P16 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P17 | c.2179G>A | p.Val727Met | c.1864G>A | p.Ala622Thr | K+K | Reported | Reported | LP | LP | CH |
| P18 | c.1853T>C | p.Ile618Thr | c.1853T>C | p.Ile618Thr | K+K | Reported | Reported | P | P | HM |
| P19 | c.2179G>A | p.Val727Met | c.1003G>A | p.Gly335Arg | K+E | Reported | Reported | LP | LP | CH |
| P20 | c.2179G>A | p.Val727Met | c.1174T>C | p.Tyr392His | K+E | Reported | Reported | LP | LP | CH |
| P21 | c.2179G>A | p.Val727Met | c.986T>C | p.Ile329Thr | K+E | Reported | Reported | LP | LP | CH |
| P22 | c.2179G>A | p.Val727Met | c.214C>T\* | p.Leu72Phe | K+E | Reported | Novel | LP | LP | CH |
| P23 | c.2179G>A | p.Val727Met | c.2189C>T | p.Ser730Leu | K+K | Reported | Reported | LP | LP | CH |
| P24 | c.1981G>A | p.Ala661Thr | c.1981G>A | p.Ala661Thr | K+K | Reported | Reported | LP | LP | HM |
| P25 | c.2179G>A | p.Val727Met | c.986T>C | p.Ile329Thr | K+E | Reported | Reported | LP | LP | CH |
| P26 | c.2179G>A | p.Val727Met | c.1351C>T | p.Arg451Ter | K+K | Reported | Reported | LP | P | CH |
| P27 | c.2179G>A | p.Val727Met | c.1313dup | p.Ser439LysfsTer6 | K+K | Reported | Novel | LP | P | CH |
| P28 | c.2179G>A | p.Val727Met | c.2058\_2059del | p.Ile687ProfsTer47 | K+K | Reported | Novel | LP | P | CH |
| P29 | c.2179G>A | p.Val727Met | c.2196G>C\* | p.Leu732Phe | K+K | Reported | Novel | LP | LP | CH |
| P30 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P31 | c.2179G>A | p.Val727Met | c.477C>G | p.Ile159Met | K+E | Reported | Novel | LP | LP | CH |
| P32 | c.2179G>A | p.Val727Met | c.478C>T | p.Arg160Ter | K+E | Reported | Reported | LP | P | CH |
| P33 | c.2179G>A | p.Val727Met | c.268C>T | p.Arg90Ter | K+E | Reported | Reported | LP | P | CH |
| P34 | c.2179G>A | p.Val727Met | c.880C>T | p.Arg294Ter | K+E | Reported | Reported | LP | P | CH |
| P35 | c.971A>G | p.His324Arg | c.971A>G | p.His324Arg | E+E | Reported | Reported | LP | LP | HM |
| P36 | c.2179G>A | p.Val727Met | c.1664C>T | p.Ala555Val | K+K | Reported | Reported | LP | P | CH |
| P37 | c.2179G>A | p.Val727Met | c.1748A>G | p.His583Arg | K+K | Reported | Novel | LP | LP | CH |
| P38 | c.2179G>A | p.Val727Met | c.729\_738delAGATTACATT | p.Asp244LeufsTer7 | K+E | Reported | Novel | LP | P | CH |
| P39 | c.1823C>G | p.Pro608Arg | c.1823C>G | p.Pro608Arg | K+K | Reported | Reported | LP | LP | HM |
| P40 | c.2179G>A | p.Val727Met | c.2098G>A | p.Gly700Arg | K+K | Reported | Reported | LP | LP | CH |
| P41 | c.2179G>A | p.Val727Met | c.2005G>A | p.Ala669Thr | K+K | Reported | Novel | LP | LP | CH |
| P42 | c.2179G>A | p.Val727Met | c.2005G>A | p.Ala669Thr | K+K | Reported | Novel | LP | LP | CH |
| P43 | c.2179G>A | p.Val727Met | c.384T>G\* | p.Asp128Glu | K+E | Reported | Novel | LP | LP | CH |
| P44 | c.2179G>A | p.Val727Met | c.533G>A | p.Arg178Lys | K+E | Reported | Novel | LP | LP | CH |
| P45 | c.2179G>A | p.Val727Met | c.723del | p.Lys241AsnfsTer13 | K+E | Reported | Novel | LP | P | CH |
| P46 | c.2179G>A | p.Val727Met | c.97G>T | p.Glu33Ter | K+E | Reported | Reported | LP | P | CH |
| P47 | c.2179G>A | p.Val727Met | c.1505G>A | p.Gly502Asp | K+K | Reported | Reported | LP | LP | CH |
| P48 | c.2179G>A | p.Val727Met | c. 986T>C | p.Ile329Thr | K+E | Reported | Reported | LP | LP | CH |
| P49 | c.2179G>A | p.Val727Met | c.1174T>C | p.Tyr392His | K+E | Reported | Reported | LP | LP | CH |
| P50 | c.2179G>A | p.Val727Met | c.1778\_1780del | p.Phe593del | K+K | Reported | Novel | LP | LP | CH |
| P51 | c.2179G>A | p.Val727Met | c.2098G>A | p.Gly700Arg | K+K | Reported | Reported | LP | LP | CH |
| P52 | c.2179G>A | p.Val727Met | c.752A>C | p.His251Pro | K+E | Reported | Novel | LP | LP | CH |
| P53 | c.2179G>A | p.Val727Met | c.2058\_2059del | p.Ile687ProfsTer47 | K+K | Reported | Novel | LP | P | CH |
| P54 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P55 | c.2179G>A | p.Val727Met | c.1783G>A | p.Ala595Thr | K+K | Reported | Novel | LP | LP | CH |
| P56 | c.2179G>A | p.Val727Met | c.1368C>G\* | p.Ser456Arg | K+K | Reported | Novel | LP | LP | CH |
| P57 | c.2179G>A | p.Val727Met | c.1474\_1475insCAGCAGAATCTG | p.Ala491\_Val492insAlaAlaGluSer | K+K | Reported | Novel | LP | LP | CH |
| P58 | c.971A>G | p.His324Arg | c.971A>G | p.His324Arg | E+E | Reported | Reported | LP | LP | HM |
| P59 | c.2179G>A | p.Val727Met | c.1153C>T | p.Gln385Ter | K+E | Reported | Novel | LP | P | CH |
| P60 | c.2179G>A | p.Val727Met | c.1796G>T | p.Gly599Val | K+K | Reported | Reported | LP | LP | CH |
| P61 | c.1853T>C | p.Ile618Thr | c.1853T>C | p.Ile618Thr | K+K | Reported | Reported | P | P | HM |
| P62 | c.2179G>A | p.Val727Met | c.1009C>T | p.Arg337Ter | K+E | Reported | Reported | LP | P | CH |
| P63 | c.2179G>A | p.Val727Met | c.925\_927delinsTTGGGAT | p.Ala309LeufsTer4 | K+E | Reported | Novel | LP | P | CH |
| P64 | c.2179G>A | p.Val727Met | c.1664C>T | p.Ala555Val | K+K | Reported | Reported | LP | P | CH |
| P65 | c.2179G>A | p.Val727Met | c.2179G>A | p.Val727Met | K+K | Reported | Reported | LP | LP | HM |
| P66 | c.2179G>A | p.Val727Met | c.478C>T | p.Arg160Ter | K+E | Reported | Reported | LP | P | CH |
| P67 | c.2179G>A | p.Val727Met | c.880C>T | p.Arg294Ter | K+E | Reported | Reported | LP | P | CH |
| P68 | c.2179G>A | p.Val727Met | c.1853T>C | p.Ile618Thr | K+K | Reported | Reported | LP | P | CH |
| P69 | c.1981G>A | p.Ala661Thr | c.1981G>A | p.Ala661Thr | K+K | Reported | Reported | LP | LP | HM |
| P70 | c.2179G>A | p.Val727Met | c.657A>C | p.Lys219Asn | K+E | Reported | Novel | LP | LP | CH |
| P71 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P72 | c.2179G>A | p.Val727Met | c.1154\_1157dup | p.Tyr386Ter | K+E | Reported | Novel | LP | P | CH |
| P73 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P74 | c.1853T>C | p.Ile618Thr | c.1853T>C | p.Ile618Thr | K+K | Reported | Reported | P | P | HM |
| P75 | c.2179G>A | p.Val727Met | c.1919T>G | p.Leu640Arg | K+K | Reported | Novel | LP | LP | CH |
| P76 | c.2179G>A | p.Val727Met | c.124C>T | p.Arg42Trp | K+E | Reported | Reported | LP | LP | CH |
| P77 | c.2179G>A | p.Val727Met | c.1632G>A | p.Trp544Ter | K+K | Reported | Reported | LP | P | CH |
| P78 | c.922C>T | p.Arg308Cys | c.922C>T | p.Arg308Cys | E+E | Reported | Reported | LP | LP | HM |
| P79 | c.2196G>C\* | p.Leu732Phe | c.2196G>C\* | p.Leu732Phe | K+K | Novel | Novel | LP | LP | HM |
| P80 | c.971A>G | p.His324Arg | c.971A>G | p.His324Arg | E+E | Reported | Reported | LP | LP | HM |
| P81 | c.2179G>A | p.Val727Met | c.880C>T | p.Arg294Ter | K+E | Reported | Reported | LP | P | CH |
| P82 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P83 | c.2179G>A | p.Val727Met | c.1251del | p.Lys417AsnfsTer7 | K+K | Reported | Reported | LP | P | CH |
| P84 | c.2179G>A | p.Val727Met | c.1709C>A\* | p.Thr570Lys | K+K | Reported | Novel | LP | LP | CH |
| P85 | c.2179G>A | p.Val727Met | c.829C>T | p.Arg277Trp | K+E | Reported | Reported | LP | P | CH |
| P86 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P87 | c.2179G>A | p.Val727Met | c.1664C>T | p.Ala555Val | K+K | Reported | Reported | LP | P | CH |
| P88 | c.2179G>A | p.Val727Met | c.2189C>T | p.Ser730Leu | K+K | Reported | Reported | LP | LP | CH |
| P89 | c.2179G>A | p.Val727Met | c.52-3C>G | 3 splice site | K | Reported | Novel | LP | VUS | CH |
| P90 | c.2179G>A | p.Val727Met | c.544A>G | p.Thr182Ala | K+E | Reported | Novel | LP | LP | CH |
| P91 | c.2179G>A | p.Val727Met | c.1451G>A | p.Cys484Tyr | K+K | Reported | Novel | LP | LP | CH |
| P92 | c.2179G>A | p.Val727Met | c.1054C>T | p. Arg352Cys | K+E | Reported | Reported | LP | LP | CH |
| P93 | c.2179G>A | p.Val727Met | c.115C>T | p.Arg39Ter | K+E | Reported | Reported | LP | P | CH |
| P94 | c.2179G>A | p.Val727Met | c.173C>T | p.Pro58Leu | K+E | Reported | Reported | LP | LP | CH |
| P95 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P96 | c.196G>A | p.Glu66Lys | c.196G>A | p.Glu66Lys | E+E | Reported | Reported | LP | LP | HM |
| P97 | c.2179G>A | p.Val727Met | c.1748A>G | p.His583Arg | K+K | Reported | Novel | LP | P | CH |
| P98 | c.2179G>A | p.Val727Met | c.1685G>A | p.Gly562Glu | K+K | Reported | Novel | LP | LP | CH |
| P99 | c.2179G>A | p.Val727Met | c.1368C>G\* | p.Ser456Arg | K+K | Reported | Novel | LP | LP | CH |
| P100 | c.2179G>A | p.Val727Met | c.1796G>T | p.Gly599Val | K+K | Reported | Reported | LP | LP | CH |
| P101 | c.1981G>A | p.Ala661Thr | c.1981G>A | p.Ala661Thr | K+K | Reported | Reported | LP | LP | HT |
| P102 | c.2179G>A | p.Val727Met | c.1075+2T>C | - | K+E | Reported | Novel | LP | LP | CH |
| P103 | c.2179G>A | p.Val727Met | c.562C>A | p.His188Asn | K+E | Reported | Reported | LP | LP | CH |
| P104 | c.1837G>A | p.Gly613Arg | c.1837G>A | p.Gly613Arg | K+K | Novel | Novel | LP | LP | HM |
| P105 | c.2179G>A | p.Val727Met | c.1111C>T | p.Gln371Ter | K+E | Reported | Novel | LP | P | CH |
| P106 | c.2179G>A | p.Val727Met | c.830G>A | p.Arg277Gln | K+E | Reported | Reported | LP | P | CH |
| P107 | c.2179G>A | p.Val727Met | c.1505G>A | p.Gly502Asp | K+K | Reported | Reported | LP | LP | CH |
| P108 | c.2179G>A | p.Val727Met | c.1374+1\_1374+2del | - | K+K | Reported | Novel | LP | P | CH |
| P109 | c.2179G>A | p.Val727Met | c.263C>T | p.Thr88Ile | K+E | Reported | Novel | LP | LP | CH |
| P110 | c.2179G>A | p.Val727Met | c.1664C>T | p.Ala555Val | K+K | Reported | Reported | LP | P | CH |
| P111 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P112 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P113 | c.2179G>A | p.Val727Met | c.2062C>T | p.Leu688Phe | K+K | Reported | Novel | LP | LP | CH |
| P114 | c.2179G>A | p.Val727Met | c.559T>C | p.Tyr187His | K+E | Reported | Reported | LP | LP | CH |
| P115 | c.2179G>A | p.Val727Met | c.1374+1\_1374+2del | - | K+K | Reported | Novel | LP | P | CH |
| P116 | c.2179G>A | p.Val727Met | c.1823C>G | p.Pro680Arg | K+K | Reported | Reported | LP | LP | CH |
| P117 | c.2179G>A | p.Val727Met | c.1251del | p.Lys417AsnfsTer7 | K+K | Reported | Reported | LP | P | CH |
| P118 | c.2179G>A | p.Val727Met | c.1251del | p.Lys417AsnfsTer7 | K+K | Reported | Reported | LP | P | CH |
| P119 | c.1853T>C | p.Ile618Thr | c.1853T>C | p.Ile618Thr | K+K | Reported | Reported | P | P | HM |
| P120 | c.2179G>A | p.Val727Met | c.268C>T | p.Arg90Ter | K+E | Reported | Reported | LP | P | CH |
| P121 | c.2179G>A | p.Val727Met | c.1748A>G | p.His583Arg | K+K | Reported | Novel | LP | LP | CH |
| P122 | c.2179G>A | p.Val727Met | c.1796G>T | p.Gly599Val | K+K | Reported | Reported | LP | LP | CH |
| P123 | c.2179G>A | p.Val727Met | c.1796G>T | p.Gly599Val | K+K | Reported | Reported | LP | LP | CH |
| P124 | c.2179G>A | p.Val727Met | c.2179G>A | p.Val727Met | K+K | Reported | Reported | LP | LP | HM |
| P125 | c.830G>A | p.Arg277Gln | c.425G>T | p.Gly142Val | E+E | Reported | Novel | P | LP | CH |
| P126 | c.2179G>A | p.Val727Met | c.215T>G | p.Leu72Arg | K+E | Reported | Novel | LP | LP | CH |
| P127 | c.1837G>A | p.Gly613Arg | c.1837G>A | p.Gly613Arg | K+K | Novel | Novel | LP | LP | HM |
| P128 | c.2179G>A | p.Val727Met | c.1505G>A | p.Gly502Asp | K+K | Reported | Reported | LP | LP | CH |
| P129 | c.2179G>A | p.Val727Met | c.1578G>A | p.Trp526Ter | K+K | Reported | Reported | LP | P | CH |
| P130 | c.2179G>A | p.Val727Met | c.1174T>C | p.Tyr392His | K+K | Reported | Reported | LP | LP | CH |
| P131 | c.1853T>C | p.Ile618Thr | c.1853T>C | p.Ile618Thr | K+K | Reported | Reported | P | P | HM |
| P132 | c.2179G>A | p.Val727Met | c.445G>A | p.Ala149Thr | K+E | Reported | Novel | LP | LP | CH |
| P133 | c.2179G>A | p.Val727Met | c.258-8G>A | 3 splice site | K | Reported | Reported | LP | VUS | CH |
| P134 | c.2179G>A | p.Val727Met | c.1351C>T | p.Arg451Ter | K+K | Reported | Reported | LP | P | CH |
| P135 | c.2179G>A | p.Val727Met | c.1006del | p.Val336PhefsTer11 | K+E | Reported | Novel | LP | P | CH |
| P136 | c.2179G>A | p.Val727Met | c.1909+2T>C | - | K | Reported | Novel | LP | LP | CH |
| P137 | c.2179G>A | p.Val727Met | c.533G>A | p.Arg178Lys | K+E | Reported | Novel | LP | LP | CH |
| P138 | c.1823C>G | p.Pro608Arg | c.1823C>G | p.Pro608Arg | K+K | Reported | Reported | LP | LP | HM |
| P139 | c.2179G>A | p.Val727Met | c.2098G>A | p.Gly700Arg | K+K | Reported | Reported | LP | LP | CH |
| P140 | c.2179G>A | p.Val727Met | c.115C>T | p.Arg39Ter | K+E | Reported | Reported | LP | P | CH |
| P141 | c.2179G>A | p.Val727Met | c.173C>T | p.Pro58Leu | K+E | Reported | Reported | LP | LP | CH |
| P142 | c.2179G>A | p.Val727Met | c.748C>T | p.Gln250Ter | K+E | Reported | Reported | LP | P | CH |
| P143 | c.2179G>A | p.Val727Met | c.18989delA | p.Lys633SerfsTer41 | K+K | Reported | Novel | P | LP | CH |
| P144 | c.2179G>A | p.Val727Met | c.575C>T | p.Thr192Ile | K+E | Reported | Novel | LP | LP | CH |
| P145 | c.2179G>A | p.Val727Met | c.1006delG | p.Val336PhefsTer11 | K+E | Reported | Novel | LP | P | CH |
| P146 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P147 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | LP | - | HT |
| P148 | c.2179G>A | p.Val727Met | c.115C>T | p.Arg39Ter | K+E | Reported | Reported | LP | P | CH |
| P149 | c.1853T>C | p.Ile618Thr | c.1853T>C | p.Ile618Thr | K+E | Reported | Reported | P | P | HM |
| P150 | c.2179G>A | p.Val727 Met | c.516\_519dup | p.Ile173HisfsTer3 | K+E | Reported | Novel | P | P | CH |
| P151 | c.2179G>A | p.Val727Met | c.1727-2A>G | - | K | Reported | Novel | P | P | CH |
| P152 | c.1853T>C | p.Ile618Thr | c.1853T>C | p.Ile618Thr | K+K | Reported | Reported | LP | LP | HM |
| P153 | c.2179G>A | p.Val727 Met | c.1748A>G | p.His583Arg | K+K | Reported | Reported | LP | VUS | CH |
| P154 | c.1089T>A | p.Asp363Glu | c.1089T>A | p.Asp363Glu | K+K | Novel | Novel | P | P | HM |
| P155 | c.1760T>C | p.Ile587Thr | c.1760T>C | p.Ile587Thr | K+K | Novel | Novel | P | P | HM |
| P156 | c.2086G>A | p.Val696Met | C.1061\_1064dup | p.Tyr355Ter | K+E | Reported | Novel | P | P | CH |
| P157 | c.2179G>A | p.Val727Met | - | - | K | Reported | - | P | - | HT |

***Footnotes:*** *K – Kinase domain, E – Epimerase domain, P – Pathogenic, LP – Likely Pathogenic, VUS – Variant of Unknown Significance, CH – Compound Heterozygous, HM – Homozygous, HT - Heterozygous*