

**S1- Table A:** patient characteristics (n=10)

| Characteristics                      | Mean(SD)           |
|--------------------------------------|--------------------|
| <b>Age(Year)</b>                     | <b>50.3 (11.6)</b> |
| <b>Gender</b>                        | <b>n (%)</b>       |
| Male                                 | 6 (60)             |
| Female                               | 4 (40)             |
| <b>Histopathology</b>                |                    |
| Adenocarcinoma                       | 1 (10)             |
| Squamous cell carcinoma              | 9 (90)             |
| <b>Tumor site</b>                    |                    |
| Upper thoracic                       | 2 (20)             |
| Mid thoracic                         | 2 (20)             |
| Lower thoracic                       | 6 (60)             |
| <b>Grade of differentiation</b>      |                    |
| Well differentiated                  | 3 (30)             |
| Moderately differentiated            | 5 (50)             |
| Poorly differentiated                | 2 (20)             |
| <b>cT stage</b>                      |                    |
| T2                                   | 1 (10)             |
| T3                                   | 5 (50)             |
| T4                                   | 4 (40)             |
| <b>cN stage</b>                      |                    |
| N0                                   | 8 (80)             |
| N1                                   | 2 (20)             |
| <b>M stage</b>                       |                    |
| M0                                   | 9 (90)             |
| M1                                   | 1 (10)             |
| <b>Clinical stage</b>                |                    |
| II                                   | 6 (60)             |
| III                                  | 1 (10)             |
| IV                                   | 3 (30)             |
| <b>Tumor length</b>                  |                    |
| <5 cm                                | 3 (30)             |
| 5-10 cm                              | 6 (60)             |
| > 10 cm                              | 1 (10)             |
| <b>Surgery</b>                       |                    |
| Performed (lap McKeon Esophagectomy) | 2 (20)             |
| Not Performed                        | 8 (80)             |

**S2- Table B:** Comprehensive list of novel potentially pathogenic missense variants (n=331) categorized by gene

| Gene |                | cDNA Change (HGVS.c)                                                                      | Amino Acid Substitution (HGVS.p)                                                                               |
|------|----------------|-------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------|
| 1.   | <i>RPS4X</i>   | c.668C>T: 1, c.665T>C: 1, c.476C>T: 1, c.452A>G: 1, c.433C>T: 1, c.454C>T: 1, c.442C>T: 1 | p.Ser223Phe: 1, p.Leu222Pro: 1, p.Thr159Ile: 1, p.Asp151Gly: 1, p.Arg145Cys: 1, p.Pro152Ser: 1, p.Arg148Cys: 1 |
| 2.   | <i>PSMCI</i>   | c.1141G>C: 1, c.980T>C: 1, c.791C>T: 1, c.803G>T: 1                                       | p.Asp381His: 1, p.Val327Ala: 1, p.Pro264Leu: 1, p.Arg268Leu: 1                                                 |
| 3.   | <i>HOMER2</i>  | c.221T>C: 1, c.218A>C: 1, c.188C>A: 1, c.176G>A: 1                                        | p.Phe74Ser: 1, p.Lys73Thr: 1, p.Pro63Gln: 1, p.Ser59Asn: 1                                                     |
| 4.   | <i>PPAT</i>    | c.1049A>C: 2, c.1247G>C: 1, c.1238T>A: 1, c.1051C>T: 1                                    | p.Asn350Thr: 2, p.Arg416Pro: 1, p.Val413Glu: 1, p.Arg351Trp: 1                                                 |
| 5.   | <i>TPM3</i>    | c.317G>A: 1, c.523G>A: 1, c.722A>G: 1                                                     | p.Arg106His: 1, p.Gly175Arg: 1, p.Glu241Gly: 1                                                                 |
| 6.   | <i>RANBP9</i>  | c.1358T>G: 1, c.1355T>G: 1, c.1352A>G: 1                                                  | p.Val453Gly: 1, p.Met452Arg: 1, p.Glu451Gly: 1                                                                 |
| 7.   | <i>NALCN</i>   | c.2846T>C: 1, c.3128T>A: 1, c.3125A>T: 1                                                  | p.Val949Ala: 1, p.Leu1043Gln: 1, p.Lys1042Ile: 1                                                               |
| 8.   | <i>PTPN11</i>  | c.1493G>A: 1, c.194T>C: 1, c.197A>T: 1                                                    | p.Arg498Gln: 1, p.Leu65Pro: 1, p.Tyr66Phe: 1                                                                   |
| 9.   | <i>ARIH2</i>   | c.1280C>T: 1, c.1282T>A: 1, c.1291G>A: 1                                                  | p.Thr427Ile: 1, p.Tyr428Asn: 1, p.Ala431Thr: 1                                                                 |
| 10.  | <i>BMPRI1B</i> | c.1222A>C: 1, c.1571T>A: 1                                                                | p.Met408Leu: 1, p.Met524Lys: 1                                                                                 |
| 11.  | <i>HOXA13</i>  | c.1130A>G: 1, c.1129G>A: 1                                                                | p.Glu377Gly: 1, p.Glu377Lys: 1                                                                                 |
| 12.  | <i>PRKAA2</i>  | c.1213G>C: 1, c.1214G>T: 1                                                                | p.Gly405Arg: 1, p.Gly405Val: 1                                                                                 |
| 13.  | <i>SCN8A</i>   | c.4496A>G: 1, c.4502A>G: 1                                                                | p.Lys1499Arg: 1, p.Gln1501Arg: 1                                                                               |
| 14.  | <i>HDAC1</i>   | c.785G>A: 1, c.1016C>A: 1                                                                 | p.Gly262Asp: 1, p.Pro339Gln: 1                                                                                 |
| 15.  | <i>LRP2</i>    | c.4058G>T: 1, c.1575T>G: 1                                                                | p.Cys1353Phe: 1, p.Phe525Leu: 1                                                                                |
| 16.  | <i>HNRNPA3</i> | c.502G>A: 1, c.523G>A: 1                                                                  | p.Gly168Arg: 1, p.Asp175Asn: 1                                                                                 |
| 17.  | <i>FECH</i>    | c.856G>T: 1, c.847C>T: 1                                                                  | p.Val286Leu: 1, p.Pro283Ser: 1                                                                                 |
| 18.  | <i>PSMC5</i>   | c.215A>G: 1, c.220G>A: 1                                                                  | p.Tyr72Cys: 1, p.Gly74Arg: 1                                                                                   |
| 19.  | <i>MTMR2</i>   | c.554C>T: 1, c.548C>G: 1                                                                  | p.Pro185Leu: 1, p.Ala183Gly: 1                                                                                 |
| 20.  | <i>ABCB1</i>   | c.1735G>A: 1, c.1730G>C: 1                                                                | p.Gly579Ser: 1, p.Arg577Thr: 1                                                                                 |
| 21.  | <i>RPL23</i>   | c.23G>A: 1, c.16C>T: 1                                                                    | p.Gly8Glu: 1, p.Arg6Cys: 1                                                                                     |
| 22.  | <i>ITPRI</i>   | c.7232T>A: 1, c.7241T>G: 1                                                                | p.Phe2411Tyr: 1, p.Val2414Gly: 1                                                                               |
| 23.  | <i>COL5A2</i>  | c.1133G>T: 1, c.3001C>G: 1                                                                | p.Gly378Val: 1, p.Arg1001Gly: 1                                                                                |
| 24.  | <i>TBL1XR1</i> | c.1451G>A: 1, c.1439A>G: 1                                                                | p.Gly484Asp: 1, p.Tyr480Cys: 1                                                                                 |
| 25.  | <i>ITGB1</i>   | c.128C>A: 1, c.127C>A: 1                                                                  | p.Pro43Gln: 1, p.Pro43Thr: 1                                                                                   |
| 26.  | <i>ZFAND1</i>  | c.235T>C: 1, c.205T>C: 1                                                                  | p.Cys79Arg: 1, p.Cys69Arg: 1                                                                                   |
| 27.  | <i>PBX2</i>    | c.896G>A: 1, c.880T>C: 1                                                                  | p.Arg299Lys: 1, p.Trp294Arg: 1                                                                                 |

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| 28. | <i>TCP1</i>    | c.1489G>C: 1, c.1034T>C: 1 | p.Asp497His: 1, p.Leu345Ser: 1   |
| 29. | <i>HNRNPD</i>  | c.686C>T: 1, c.675C>G: 1   | p.Thr229Ile: 1, p.Phe225Leu: 1   |
| 30. | <i>CALM2</i>   | c.470T>C: 1, c.464G>A: 1   | p.Val157Ala: 1, p.Arg155His: 1   |
| 31. | <i>PEX5</i>    | c.428T>G: 1, c.429T>A: 1   | p.Phe143Cys: 1, p.Phe143Leu: 1   |
| 32. | <i>PCBD2</i>   | c.322G>A: 1, c.338C>T: 1   | p.Asp108Asn: 1, p.Thr113Ile: 1   |
| 33. | <i>CACNA1E</i> | c.4182C>G: 1, c.3626G>A: 1 | p.Ser1394Arg: 1, p.Gly1209Asp: 1 |
| 34. | <i>FOXO1</i>   | c.1956G>C: 1, c.1943C>G: 1 | p.Trp652Cys: 1, p.Thr648Arg: 1   |
| 35. | <i>GPD2</i>    | c.443T>C: 1, c.451G>T: 1   | p.Leu148Pro: 1, p.Ala151Ser: 1   |
| 36. | <i>KARS</i>    | c.1391C>T: 1, c.1803G>A: 1 | p.Pro464Leu: 1, p.Met601Ile: 1   |
| 37. | <i>ABCC2</i>   | c.4186G>T: 1, c.4187A>C: 1 | p.Asp1396Tyr: 1, p.Asp1396Ala: 1 |
| 38. | <i>AFG3L2</i>  | c.1364G>A: 2               | p.Arg455Gln: 2                   |
| 39. | <i>CHCHD2</i>  | c.350A>G: 2                | p.Glu117Gly: 2                   |
| 40. | <i>HNRNPA1</i> | c.547A>C: 2                | p.Lys183Gln: 2                   |
| 41. | <i>RYR3</i>    | c.8318C>A: 1               | p.Thr2773Asn: 1                  |
| 42. | <i>STAT3</i>   | c.1870G>T: 1               | p.Val624Leu: 1                   |
| 43. | <i>ZNF827</i>  | c.2515C>T: 1               | p.His839Tyr: 1                   |
| 44. | <i>GRIK1</i>   | c.1172A>T: 1               | p.Asp391Val: 1                   |
| 45. | <i>STK4</i>    | c.218A>G: 1                | p.Glu73Gly: 1                    |
| 46. | <i>CDH10</i>   | c.1981G>T: 1               | p.Gly661Cys: 1                   |
| 47. | <i>MOCS1</i>   | c.905G>T: 1                | p.Ser302Ile: 1                   |
| 48. | <i>KCNQ5</i>   | c.637T>C: 1                | p.Ser213Pro: 1                   |
| 49. | <i>RBL1</i>    | c.1657A>G: 1               | p.Lys553Glu: 1                   |
| 50. | <i>ASZ1</i>    | c.603G>T: 1                | p.Leu201Phe: 1                   |
| 51. | <i>PAX5</i>    | c.74G>A: 1                 | p.Gly25Glu: 1                    |
| 52. | <i>YME1L1</i>  | c.1700T>C: 1               | p.Met567Thr: 1                   |
| 53. | <i>ARMC4</i>   | c.2865T>A: 1               | p.Asn955Lys: 1                   |
| 54. | <i>PRRG4</i>   | c.194T>C: 1                | p.Leu65Pro: 1                    |
| 55. | <i>KLF8</i>    | c.985C>T: 1                | p.His329Tyr: 1                   |
| 56. | <i>CAT</i>     | c.451C>G: 1                | p.Pro151Ala: 1                   |
| 57. | <i>PHB</i>     | c.415C>G: 1                | p.Leu139Val: 1                   |
| 58. | <i>LALBA</i>   | c.275G>A: 1                | p.Cys92Tyr: 1                    |
| 59. | <i>CD63</i>    | c.494T>G: 1                | p.Val165Gly: 1                   |
| 60. | <i>RCBTB1</i>  | c.598G>T: 1                | p.Gly200Cys: 1                   |
| 61. | <i>KLHL1</i>   | c.710T>C: 1                | p.Phe237Ser: 1                   |
| 62. | <i>AQR</i>     | c.3775A>G: 1               | p.Thr1259Ala: 1                  |

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| 63. | <i>ABCD2</i>   | c.1885T>G: 1 | p.Tyr629Asp: 1  |
| 64. | <i>OCRL</i>    | c.1487C>A: 1 | p.Pro496Gln: 1  |
| 65. | <i>COL4A5</i>  | c.1625G>A: 1 | p.Gly542Glu: 1  |
| 66. | <i>SLC4A8</i>  | c.2866A>C: 1 | p.Thr956Pro: 1  |
| 67. | <i>PIK3C2A</i> | c.4169T>A: 1 | p.Ile1390Asn: 1 |
| 68. | <i>HBP1</i>    | c.995G>A: 1  | p.Gly332Asp: 1  |
| 69. | <i>HSP90B1</i> | c.249C>A: 1  | p.Asn83Lys: 1   |
| 70. | <i>KCNH5</i>   | c.1540T>C: 1 | p.Ser514Pro: 1  |
| 71. | <i>ESR1</i>    | c.1519G>C: 1 | p.Ala507Pro: 1  |
| 72. | <i>HERC3</i>   | c.73C>A: 1   | p.Pro25Thr: 1   |
| 73. | <i>INO80</i>   | c.1061A>G: 1 | p.Glu354Gly: 1  |
| 74. | <i>TRPM7</i>   | c.2114C>A: 1 | p.Ala705Asp: 1  |
| 75. | <i>COL8A1</i>  | c.941G>C: 1  | p.Gly314Ala: 1  |
| 76. | <i>TAOK1</i>   | c.998A>T: 1  | p.Glu333Val: 1  |
| 77. | <i>CLCN7</i>   | c.1983C>G: 1 | p.Phe661Leu: 1  |
| 78. | <i>GNAO1</i>   | c.448A>C: 1  | p.Asn150His: 1  |
| 79. | <i>ZFP1</i>    | c.1145G>A: 1 | p.Cys382Tyr: 1  |
| 80. | <i>AQP4</i>    | c.568G>A: 1  | p.Ala190Thr: 1  |
| 81. | <i>RPL11</i>   | c.80G>A: 1   | p.Gly27Glu: 1   |
| 82. | <i>ITGB6</i>   | c.466G>A: 1  | p.Gly156Ser: 1  |
| 83. | <i>ATP2B2</i>  | c.884A>T: 1  | p.Glu295Val: 1  |
| 84. | <i>ABCB10</i>  | c.1513C>G: 1 | p.Pro505Ala: 1  |
| 85. | <i>USH2A</i>   | c.2738G>T: 1 | p.Cys913Phe: 1  |
| 86. | <i>DDR2</i>    | c.319G>C: 1  | p.Ala107Pro: 1  |
| 87. | <i>STT3B</i>   | c.1887C>A: 1 | p.Ser629Arg: 1  |
| 88. | <i>COL11A1</i> | c.2249A>G: 1 | p.Glu750Gly: 1  |
| 89. | <i>COL25A1</i> | c.1418G>A: 1 | p.Gly473Glu: 1  |
| 90. | <i>TRPV1</i>   | c.1667C>T: 1 | p.Thr556Ile: 1  |
| 91. | <i>HADH</i>    | c.823C>T: 1  | p.Pro275Ser: 1  |
| 92. | <i>TASPI</i>   | c.149C>T: 1  | p.Ala50Val: 1   |
| 93. | <i>CLDN10</i>  | c.157T>G: 1  | p.Cys53Gly: 1   |
| 94. | <i>AMT</i>     | c.779C>T: 1  | p.Ala260Val: 1  |
| 95. | <i>SOD3</i>    | c.461G>A: 1  | p.Gly154Asp: 1  |
| 96. | <i>HAND2</i>   | c.419C>T: 1  | p.Thr140Ile: 1  |
| 97. | <i>BMP6</i>    | c.1352C>T: 1 | p.Ala451Val: 1  |

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| 98.  | <i>RRM2B</i>    | c.793G>T: 1  | p.Val265Leu: 1  |
| 99.  | <i>ABCA1</i>    | c.5882G>A: 1 | p.Gly1961Glu: 1 |
| 100. | <i>ITGA8</i>    | c.2034T>A: 1 | p.Asn678Lys: 1  |
| 101. | <i>CNNM1</i>    | c.2042T>C: 1 | p.Val681Ala: 1  |
| 102. | <i>COX15</i>    | c.971T>A: 1  | p.Phe324Tyr: 1  |
| 103. | <i>CHRNA10</i>  | c.1070G>A: 1 | p.Cys357Tyr: 1  |
| 104. | <i>AMPD3</i>    | c.1415A>G: 1 | p.Tyr472Cys: 1  |
| 105. | <i>TUBA1C</i>   | c.1034T>C: 1 | p.Val345Ala: 1  |
| 106. | <i>GPR12</i>    | c.412C>T: 1  | p.Arg138Cys: 1  |
| 107. | <i>RASA3</i>    | c.2270A>G: 1 | p.Tyr757Cys: 1  |
| 108. | <i>ORC4</i>     | c.200G>T: 1  | p.Gly67Val: 1   |
| 109. | <i>BAZ1A</i>    | c.1660A>G: 1 | p.Arg554Gly: 1  |
| 110. | <i>SPATA5L1</i> | c.1484T>C: 1 | p.Leu495Pro: 1  |
| 111. | <i>ISL2</i>     | c.328T>C: 1  | p.Tyr110His: 1  |
| 112. | <i>SETD1A</i>   | c.4877G>T: 1 | p.Arg1626Leu: 1 |
| 113. | <i>NXN</i>      | c.613T>G: 1  | p.Cys205Gly: 1  |
| 114. | <i>NF1</i>      | c.4349C>A: 1 | p.Ala1450Asp: 1 |
| 115. | <i>CCT6B</i>    | c.1451G>C: 1 | p.Gly484Ala: 1  |
| 116. | <i>SCN4A</i>    | c.689T>C: 1  | p.Ile230Thr: 1  |
| 117. | <i>SMARCA4</i>  | c.3404G>A: 1 | p.Arg1135Gln: 1 |
| 118. | <i>IZUMO1</i>   | c.698G>A: 1  | p.Cys233Tyr: 1  |
| 119. | <i>TRIM28</i>   | c.626G>A: 1  | p.Cys209Tyr: 1  |
| 120. | <i>STAU1</i>    | c.710G>A: 1  | p.Gly237Glu: 1  |
| 121. | <i>MOV10L1</i>  | c.713G>T: 1  | p.Arg238Met: 1  |
| 122. | <i>ALPI</i>     | c.1457G>A: 1 | p.Cys486Tyr: 1  |
| 123. | <i>SLC20A1</i>  | c.191T>C: 1  | p.Ile64Thr: 1   |
| 124. | <i>GPM6B</i>    | c.848T>C: 1  | p.Leu283Pro: 1  |
| 125. | <i>HMBOX1</i>   | c.803G>A: 1  | p.Arg268Gln: 1  |
| 126. | <i>PEX10</i>    | c.112G>A: 1  | p.Gly38Ser: 1   |
| 127. | <i>IRF6</i>     | c.1127G>A: 1 | p.Gly376Glu: 1  |
| 128. | <i>COL3A1</i>   | c.1177G>A: 1 | p.Gly393Ser: 1  |
| 129. | <i>COL6A3</i>   | c.6185G>T: 1 | p.Gly2062Val: 1 |
| 130. | <i>RAD54L2</i>  | c.920T>A: 1  | p.Met307Lys: 1  |
| 131. | <i>ALAS1</i>    | c.965C>T: 1  | p.Ala322Val: 1  |
| 132. | <i>TSPAN5</i>   | c.544G>T: 1  | p.Val182Phe: 1  |

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| 133. | <i>KRT16</i>       | c.1217T>C: 1 | p.Val406Ala: 1  |
| 134. | <i>TRIO</i>        | c.647T>A: 1  | p.Ile216Asn: 1  |
| 135. | <i>SERPINB9</i>    | c.935C>T: 1  | p.Ser312Phe: 1  |
| 136. | <i>RPS10-NUDT3</i> | c.239G>A: 1  | p.Arg80His: 1   |
| 137. | <i>RWDD1</i>       | c.182C>A: 1  | p.Pro61Gln: 1   |
| 138. | <i>PRSSI</i>       | c.592G>C: 1  | p.Gly198Arg: 1  |
| 139. | <i>ODF1</i>        | c.460C>G: 1  | p.Arg154Gly: 1  |
| 140. | <i>IRF2BP2</i>     | c.1649C>A: 1 | p.Pro550His: 1  |
| 141. | <i>MEGF9</i>       | c.1111T>C: 1 | p.Cys371Arg: 1  |
| 142. | <i>VIM</i>         | c.871T>A: 1  | p.Tyr291Asn: 1  |
| 143. | <i>CUL2</i>        | c.1709G>C: 1 | p.Arg570Thr: 1  |
| 144. | <i>RBM4B</i>       | c.346G>T: 1  | p.Val116Leu: 1  |
| 145. | <i>ATP11A</i>      | c.2479G>A: 1 | p.Ala827Thr: 1  |
| 146. | <i>GABRG3</i>      | c.773T>C: 1  | p.Ile258Thr: 1  |
| 147. | <i>ABCC12</i>      | c.1610T>G: 1 | p.Val537Gly: 1  |
| 148. | <i>DSC1</i>        | c.1406C>A: 1 | p.Pro469His: 1  |
| 149. | <i>LIG1</i>        | c.1876G>T: 1 | p.Asp626Tyr: 1  |
| 150. | <i>TNFRSF4</i>     | c.190T>C: 1  | p.Cys64Arg: 1   |
| 151. | <i>LRRC47</i>      | c.998G>T: 1  | p.Arg333Leu: 1  |
| 152. | <i>KDM1A</i>       | c.1009G>T: 1 | p.Val337Phe: 1  |
| 153. | <i>CHRNA2</i>      | c.758T>A: 1  | p.Ile253Asn: 1  |
| 154. | <i>KRT17</i>       | c.820T>C: 1  | p.Trp274Arg: 1  |
| 155. | <i>EGF</i>         | c.560T>A: 1  | p.Leu187His: 1  |
| 156. | <i>GOSR1</i>       | c.551C>T: 1  | p.Ala184Val: 1  |
| 157. | <i>ABCC11</i>      | c.3919T>C: 1 | p.Ser1307Pro: 1 |
| 158. | <i>SMCHD1</i>      | c.553A>G: 1  | p.Asn185Asp: 1  |
| 159. | <i>MORC3</i>       | c.308G>A: 1  | p.Gly103Asp: 1  |
| 160. | <i>BMX</i>         | c.1970C>A: 1 | p.Pro657His: 1  |
| 161. | <i>SMS</i>         | c.929G>T: 1  | p.Gly310Val: 1  |
| 162. | <i>RLIM</i>        | c.1718G>T: 1 | p.Cys573Phe: 1  |
| 163. | <i>UQCRH</i>       | c.158G>A: 1  | p.Cys53Tyr: 1   |
| 164. | <i>OLFML3</i>      | c.658T>C: 1  | p.Tyr220His: 1  |
| 165. | <i>MSGN1</i>       | c.525C>G: 1  | p.Ile175Met: 1  |
| 166. | <i>MDH1</i>        | c.259G>T: 1  | p.Val87Leu: 1   |
| 167. | <i>MYO1B</i>       | c.1660T>C: 1 | p.Phe554Leu: 1  |

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| 168. | <i>B3GALNT1</i> | c.544G>T: 1  | p.Asp182Tyr: 1  |
| 169. | <i>TBCCD1</i>   | c.1433A>T: 1 | p.Asp478Val: 1  |
| 170. | <i>NR2F1</i>    | c.928A>C: 1  | p.Lys310Gln: 1  |
| 171. | <i>RNF14</i>    | c.826C>A: 1  | p.Pro276Thr: 1  |
| 172. | <i>ABCB4</i>    | c.3811G>A: 1 | p.Gly1271Ser: 1 |
| 173. | <i>PPP2CB</i>   | c.715C>A: 1  | p.Arg239Ser: 1  |
| 174. | <i>FXN</i>      | c.553C>G: 1  | p.Leu185Val: 1  |
| 175. | <i>GNAQ</i>     | c.282G>A: 1  | p.Met94Ile: 1   |
| 176. | <i>ACCSL</i>    | c.1498G>C: 1 | p.Glu500Gln: 1  |
| 177. | <i>SDHD</i>     | c.370G>C: 1  | p.Ala124Pro: 1  |
| 178. | <i>CHD4</i>     | c.5599G>A: 1 | p.Asp1867Asn: 1 |
| 179. | <i>SLC25A3</i>  | c.685C>G: 1  | p.Pro229Ala: 1  |
| 180. | <i>KMT5A</i>    | c.887T>C: 1  | p.Leu296Pro: 1  |
| 181. | <i>SPRED1</i>   | c.973C>G: 1  | p.Arg325Gly: 1  |
| 182. | <i>SDF2</i>     | c.265G>T: 1  | p.Gly89Cys: 1   |
| 183. | <i>MYL12A</i>   | c.362G>A: 1  | p.Gly121Asp: 1  |
| 184. | <i>KRT19</i>    | c.1142G>T: 1 | p.Arg381Leu: 1  |
| 185. | <i>VPS13C</i>   | c.201G>T: 1  | p.Leu67Phe: 1   |
| 186. | <i>ITGA11</i>   | c.3210C>G: 1 | p.Cys1070Trp: 1 |
| 187. | <i>EML5</i>     | c.2403G>T: 1 | p.Trp801Cys: 1  |
| 188. | <i>ZCCHC9</i>   | c.661C>T: 1  | p.His221Tyr: 1  |
| 189. | <i>GDI2</i>     | c.857A>G: 1  | p.Tyr286Cys: 1  |
| 190. | <i>BRCA2</i>    | c.4586G>T: 1 | p.Gly1529Val: 1 |
| 191. | <i>SLC7A8</i>   | c.550A>C: 1  | p.Thr184Pro: 1  |
| 192. | <i>ALDOA</i>    | c.230T>C: 1  | p.Ile77Thr: 1   |
| 193. | <i>DNAH9</i>    | c.6585T>A: 1 | p.Asn2195Lys: 1 |
| 194. | <i>PEX12</i>    | c.199G>C: 1  | p.Asp67His: 1   |
| 195. | <i>COL1A1</i>   | c.746C>A: 1  | p.Pro249His: 1  |
| 196. | <i>ABCD3</i>    | c.1865C>G: 1 | p.Thr622Ser: 1  |
| 197. | <i>HSD3B1</i>   | c.1057A>C: 1 | p.Thr353Pro: 1  |
| 198. | <i>EFEMP1</i>   | c.1468C>A: 1 | p.Pro490Thr: 1  |
| 199. | <i>CACNA1D</i>  | c.4296G>A: 1 | p.Met1432Ile: 1 |
| 200. | <i>KIT</i>      | c.2396T>C: 1 | p.Leu799Pro: 1  |
| 201. | <i>H2AFZ</i>    | c.341T>C: 1  | p.Ile114Thr: 1  |
| 202. | <i>SMARCA5</i>  | c.1472T>C: 1 | p.Leu491Pro: 1  |

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| 203. | <i>ABCE1</i>   | c.1099G>A: 1 | p.Gly367Arg: 1  |
| 204. | <i>F13A1</i>   | c.464T>G: 1  | p.Val155Gly: 1  |
| 205. | <i>ASCC3</i>   | c.4685T>A: 1 | p.Val1562Asp: 1 |
| 206. | <i>PLAT</i>    | c.212G>T: 1  | p.Cys71Phe: 1   |
| 207. | <i>TMEM68</i>  | c.287T>A: 1  | p.Val96Glu: 1   |
| 208. | <i>RAD54B</i>  | c.1311G>T: 1 | p.Lys437Asn: 1  |
| 209. | <i>FKTN</i>    | c.1000G>C: 1 | p.Ala334Pro: 1  |
| 210. | <i>ALG8</i>    | c.280A>C: 1  | p.Asn94His: 1   |
| 211. | <i>RAD52</i>   | c.250T>A: 1  | p.Trp84Arg: 1   |
| 212. | <i>CHD8</i>    | c.4072G>T: 1 | p.Val1358Phe: 1 |
| 213. | <i>CXADR</i>   | c.596T>A: 1  | p.Val199Glu: 1  |
| 214. | <i>PHKA1</i>   | c.588G>T: 1  | p.Glu196Asp: 1  |
| 215. | <i>MUMILI1</i> | c.1219C>T: 1 | p.Pro407Ser: 1  |
| 216. | <i>GUCY2F</i>  | c.2945G>T: 1 | p.Gly982Val: 1  |
| 217. | <i>PRCP</i>    | c.1079G>T: 1 | p.Gly360Val: 1  |
| 218. | <i>RAB30</i>   | c.131T>G: 1  | p.Val44Gly: 1   |
| 219. | <i>DYNC2H1</i> | c.9026C>A: 1 | p.Pro3009Gln: 1 |
| 220. | <i>PSMC6</i>   | c.1103C>T: 1 | p.Ala368Val: 1  |
| 221. | <i>ESCO1</i>   | c.2368T>A: 1 | p.Cys790Ser: 1  |
| 222. | <i>SLC25A5</i> | c.576C>A: 1  | p.Phe192Leu: 1  |
| 223. | <i>MTOR</i>    | c.6498G>T: 1 | p.Lys2166Asn: 1 |
| 224. | <i>KCNH1</i>   | c.1867T>C: 1 | p.Ser623Pro: 1  |
| 225. | <i>ABCA12</i>  | c.4925T>G: 1 | p.Met1642Arg: 1 |
| 226. | <i>TRPC1</i>   | c.1592G>A: 1 | p.Gly531Glu: 1  |
| 227. | <i>MME</i>     | c.2228C>A: 1 | p.Pro743Gln: 1  |
| 228. | <i>ABCC5</i>   | c.2917A>T: 1 | p.Asn973Tyr: 1  |
| 229. | <i>SCFD2</i>   | c.1895G>T: 1 | p.Gly632Val: 1  |
| 230. | <i>SLC4A4</i>  | c.1529T>A: 1 | p.Val510Asp: 1  |
| 231. | <i>MTHFD2L</i> | c.641G>A: 1  | p.Gly214Glu: 1  |
| 232. | <i>FH</i>      | c.453G>T: 1  | p.Met151Ile: 1  |
| 233. | <i>FBN2</i>    | c.2651G>T: 1 | p.Ser884Ile: 1  |
| 234. | <i>MKRN1</i>   | c.929G>C: 1  | p.Cys310Ser: 1  |
| 235. | <i>GALNT11</i> | c.317A>C: 1  | p.Asp106Ala: 1  |
| 236. | <i>RNF19A</i>  | c.1067G>T: 1 | p.Ser356Ile: 1  |
| 237. | <i>FZD6</i>    | c.554G>C: 1  | p.Cys185Ser: 1  |



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| 238. | <i>HKDC1</i>   | c.2214G>C: 1 | p.Gln738His: 1  |
| 239. | <i>OAT</i>     | c.155C>T: 1  | p.Ala52Val: 1   |
| 240. | <i>ITGB7</i>   | c.1499G>T: 1 | p.Cys500Phe: 1  |
| 241. | <i>SUOX</i>    | c.1173T>A: 1 | p.Ser391Arg: 1  |
| 242. | <i>SLC5A8</i>  | c.677G>T: 1  | p.Arg226Ile: 1  |
| 243. | <i>SYNE2</i>   | c.299T>C: 1  | p.Phe100Ser: 1  |
| 244. | <i>SMC3</i>    | c.395G>A: 1  | p.Arg132Gln: 1  |
| 245. | <i>CNTNAP2</i> | c.1707C>G: 1 | p.Cys569Trp: 1  |
| 246. | <i>GATAD1</i>  | c.340G>A: 1  | p.Gly114Arg: 1  |
| 247. | <i>HELZ</i>    | c.2537A>G: 1 | p.Asp846Gly: 1  |
| 248. | <i>ECHDC2</i>  | c.839C>A: 1  | p.Ala280Asp: 1  |
| 249. | <i>DNM3</i>    | c.416G>C: 1  | p.Gly139Ala: 1  |
| 250. | <i>TIFA</i>    | c.140T>A: 1  | p.Val47Glu: 1   |
| 251. | <i>SMAD5</i>   | c.379C>A: 1  | p.His127Asn: 1  |
| 252. | <i>ABCC10</i>  | c.2144A>G: 1 | p.Asp715Gly: 1  |
| 253. | <i>PSMC2</i>   | c.695G>A: 1  | p.Arg232Gln: 1  |
| 254. | <i>SPAG1</i>   | c.682G>A: 1  | p.Ala228Thr: 1  |
| 255. | <i>ZDHHC5</i>  | c.142C>A: 1  | p.Pro48Thr: 1   |
| 256. | <i>KRT18</i>   | c.634T>C: 1  | p.Phe212Leu: 1  |
| 257. | <i>SETDB2</i>  | c.502C>T: 1  | p.Pro168Ser: 1  |
| 258. | <i>CHRNA5</i>  | c.1019G>C: 1 | p.Arg340Pro: 1  |
| 259. | <i>MKL2</i>    | c.339G>A: 1  | p.Met113Ile: 1  |
| 260. | <i>SETBP1</i>  | c.2734A>G: 1 | p.Lys912Glu: 1  |
| 261. | <i>GRM1</i>    | c.3503A>T: 1 | p.Glu1168Val: 1 |
| 262. | <i>SLC5A3</i>  | c.2093T>A: 1 | p.Leu698Gln: 1  |
| 263. | <i>BTK</i>     | c.1322A>G: 1 | p.Glu441Gly: 1  |
| 264. | <i>CCT3</i>    | c.1064T>G: 1 | p.Ile355Ser: 1  |
| 265. | <i>F5</i>      | c.1654G>A: 1 | p.Asp552Asn: 1  |
| 266. | <i>ESRRG</i>   | c.925C>A: 1  | p.Gln309Lys: 1  |
| 267. | <i>CCT7</i>    | c.1367A>G: 1 | p.Asp456Gly: 1  |
| 268. | <i>PKP4</i>    | c.2411G>A: 1 | p.Gly804Glu: 1  |
| 269. | <i>COL4A3</i>  | c.4453C>A: 1 | p.Gln1485Lys: 1 |
| 270. | <i>PDHB</i>    | c.902T>G: 1  | p.Val301Gly: 1  |
| 271. | <i>RIPK1</i>   | c.472G>T: 1  | p.Gly158Cys: 1  |
| 272. | <i>RNF5</i>    | c.509C>T: 1  | p.Ala170Val: 1  |

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| 273. | <i>SMPDL3A</i> | c.581C>T: 1  | p.Ser194Leu: 1 |
| 274. | <i>CLCN5</i>   | c.1048C>T: 1 | p.Arg350Cys: 1 |