**Supplementary Table 2:** Truncating mutations located in the ARM2 or ARM3 domain of the APC protein.

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Gene | Variant (Human Genome Variation Society, HGVS) | Location | APC protein change | Protein domain | Family subjects | Colon polyposis phenotype | Polyp number | CRC (age at Dx) | Reference |
| *APC* | c.1557\_1561delATGCT | Exon 12 | p.(Cys520Tyrfs\*15) | ARM 2 | F1-I | AFAP | n.a. | n.a. | [67] |
| F1-II | AFAP | n.a. | n.a. |
| F1-III | AFAP | n.a. | n.a. |
| *APC* | c.1576dupA | Exon 12 | p.(Met526Asnfs\*11) | ARM 2 | F2-I | Classic FAP | >100 | n.a.. | [84] |
| *APC* | c.1586\_1587insAT | Exon 12 | p.(Val530Leufs\*5) | ARM 2 | F3-I | Classic FAP | >100 | Yes  (44) | [85] |
| *APC* | c.1594delC | Exon 12 | p.(Gln532Asnfs\*2) | ARM 2 | F4-I | Classic FAP | >100 | n.a. | [86] |
| *APC* | c.1594delC | Exon 12 | p.(Gln532Asnfs\*2) | ARM 2 | F5-I | Classic FAP | >100 | n.a. | [87] |
| *APC* | c.1602dupA | Exon 12 | p.(Ser535Ilefs\*2) | ARM 2 | F6-I | Classic FAP | >100 | n.a. | [84] |
| *APC* | c.1605\_1606delTG | Exon 12 | p.(Glu536Lysfs\*2) | ARM 2 | F7-I | Classic FAP | >100 | n.a. | [88] |
| F7-II | Classic FAP | >100 | n.a. |
| F7-III | Classic FAP | >100 | n.a. |
| *APC* | c.1609delA | Exon 12 | p.(Ser537Valfs\*12) | ARM 2/  ARM3 | F8-I | Classic FAP | >100 | n.a. | [89] |
| *APC* | c.1614dupA | Exon 12 | p.(Asp539Argfs\*21) | ARM 2/  ARM3 | F9-I | Classic FAP | >100 | n.a. | [90] |
| *APC* | c.1620dupA | Exon 12 | p.(Gln541Thrfs\*19) | ARM 2/  ARM3 | F10-I | Severe FAP | n.a. | n.a. | [91] |
| *APC* | c.1622dupA | Exon 12 | p.(Gln542Alafs\*18) | ARM 2/  ARM3 | F11-I | Classic FAP | > 100 | n.a. | [43] |
| *APC* | c.1624C>T | Exon 12 | p.Q542\* | ARM 2 | F12-I | Classic FAP | > 100 | n.a. | [53] |
| *APC* | c.1629delT | Exon 13 | p.(Ile544Leufs\*5) | ARM 3 | F13-I | Classic FAP | >100 | Yes  (42) | [92] |
| *APC* | c.1636\_1639delAGTG | Exon 13 | p.(Ser546Phefs\*2) | ARM 3 | F14-I | Classic FAP | >100 | Yes  (41) | [93] |
| *APC* | c.1642\_1643delTT | Exon 13 | p.(Leu548Glufs\*11) | ARM 3 | F15-I | Classic FAP | 100–1000 | No | [94] |
| *APC* | c.1659G>A | Exon 13 | p.W553\* | ARM 3 | F16-I | AFAP | <100 | No | [95] |
| F16-II | Classic FAP | >500 | No |
| *APC* | c.1660C>T | Exon 13 | p.R554\* | ARM 3 | F17-I | Profuse FAP | >1000 | n.a. | [96] |
| *APC* | c.1673delA | Exon 13 | p.(Asn558Ilefs\*12) | ARM 3 | F18-I | AFAP | <10 | Yes  (43) | [97] |
| *APC* | c.1699G>T | Exon 13 | p.G567\* | ARM 3 | F19-I | Classic FAP | >100 | No | [98] |
| F20-I | AFAP | <100 | n.a. | [67] |
| *APC* | c.1732G>T | Exon 13 | p.E578\* | ARM 3 | F21-I | Classic FAP | >100 | n.a. | [99] |
| *APC* | c.1682dupA | Exon 13 | p.(Thr562Aspfs\*19) | ARM 3 | F22-I | Severe FAP | n.a. | n.a. | 100] |
| F22-II | Severe FAP | n.a. | n.a. |
| F22-III | Severe FAP | n.a. | n.a. |
| F22-IV | Severe FAP | n.a. | n.a. |
| F22-V | Severe FAP | n.a. | n.a. |
| F22-VI | Severe FAP | n.a. | n.a. |
| F22-VII | Severe FAP | n.a. | n.a. |

CRC: colorectal cancer; Dx: diagnosis; n.a.: not available; Yes: presence of clinical phenotype; No: absence of clinical phenotype