|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **Patients** | **Proband** | **Age at**  **genetic test** | **APC pathogenic variant - Nucleotide HGVS** | **APC pathogenic variant - Protein HGVS** | **N. of polyps at first endoscopic exam (age)** | **Total colectomy (age)** |
| #1 | Y | 31 years | c.(?\_-85)\_(\*1\_?)del | p.(0) | colonoscopy (symptoms): 30 (11 years) | IRA (24 years) |
| #2 | Y | 25 years | c.3329C>G | p.(Ser1110\*) | colonoscopy (symptoms): 50 (25 years) | N |
| #3 | N | 9 years | c.3768dupA | p.(Glu1257Argfs\*19) | colonoscopy: > 100 (15 years) | IRA (16 years) |
| #4 | N | 8 years | c.3183\_3187delACAAA | p.(Gln1062\*) | NA | N |
| #5 | N | 8 years | c.3927\_3931delAAAGA | p.(Glu1309Aspfs\*4) | NA | N |
| #6 | N | 10 years | c.4145\_4150delTCATGTinsA | p.(Leu1382Hisfs\*2) | colonoscopy: > 100 (13 years) | N |

**Table 2: Patient FAP histories**

Legenda: Y: yes; N: no; IRA: ileorectal anastomosis; HGVS: mutation nomenclature according to the Human Genome Variation Society (HGVS; http://varnomen.hgvs.org)