**S1** Table. Pathogenic mutations found in patients’ tumors by NGS and their nucleotide changes associated with germline mutations reported from literatures.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Genes | EXON | Nucleotide change | Assumed consequence | Disease | References |
| APC | exon16 | c.4588G>T | p.E1530X | Adenomatous polyposis coli | Bisgaard,et al 1 |
|  | exon14 | c.1660C>T | p.R554X | Adenomatous polyposis coli | Fodde,et al 2 |
|  | exon16 | c.2626C>T | p.R876X | Adenomatous polyposis coli | Miyaki,et al 3 |
|  | exon16 | c.3709C>A | p.Q1237K | Adenomatous polyposis coli | Jang,et al 4 |
|  | exon8 | c.710C>G | P.S237C | Adenomatous polyposis coli | Lopez-Kostner,et al 5 |
|  | exon8 | c.646C>T | P.R216X | Adenomatous polyposis coli | Lamlum,et al 6 |
|  | exon16 | c.3340C>T | p.R1114X | Adenomatous polyposis coli | Nagase,et al 7 |
|  | exon6 | c.637C>T | p.R213X | Adenomatous polyposis coli | Miyoshi,et al 8 |
| BMPR1A | - | - | - | - | - |
| BRCA1 | exon10 | c.3710T>A | p.I1237K | Breast and/or ovarian cancer | Johannsson,et al 9 |
|  | exon20 | c.5194-2A>T |  | Breast and/or ovarian cancer | Robertson,et al 10 |
|  | exon19 | c.5252G>A | p.R1751Q | Breast and/or ovarian cancer | Gad,et al 11 |
|  | exon13 | c.6952C>T | p.R2318X | Breast and/or ovarian cancer | Zhou,et al 12 |
|  | exon10 | c.2083G>A | p.D695N | Breast and/or ovarian cancer | Greenman,et al 13 |
|  | exon10 | c.1945G>A | p.E649K | Breast and/or ovarian cancer | Li,et al 14 |
|  | exon4 | c.203T>A | p.I68K | Breast and/or ovarian cancer | Abkevich,et al 15 |
|  | exon10 | c.800C>T | p.S267L | Breast and/or ovarian cancer | Caux-Moncoutier V,et al 16 |
|  | exon6 | c.427G>A | p.E143K | Breast and/or ovarian cancer | Shattuck-Eidens,et al 17 |
|  | exon23 | c.5506G>A | p.E1836K | Breast and/or ovarian cancer | Diez-Gilbert,et al 18 |
|  | exon10 | c.2591C>A | p.S864X | Breast and/or ovarian cancer | Marroni,et al 19 |
|  | exon10 | c.1487G>A | p.R496H | Breast and/or ovarian cancer | Schoumacher,et al 20 |
|  | exon19 | c.5246C>T | p.P1749L | Breast and/or ovarian cancer | Gayther,et al 21 |
| BRCA2 | exon13 | c.6952C>T | p.R2318X | Breast and/or ovarian cancer | Wagner,et al 22 |
|  | exon24 | c.9154C>T | p.R3052W | Breast and/or ovarian cancer | Kuznetsov,et al 23 |
|  | exon11 | c.6281A>T | p.Y2094F | Breast and/or ovarian cancer | Jakubowska,et al 24 |
|  | exon9 | c.742G>A | p.A248T | Breast and/or ovarian cancer | Fackenthal,et al 25 |
|  | exon23 | c.9007G>T | p.G3003X | Breast and/or ovarian cancer | Zhang,et al 26 |
|  | exon7 | c.520C>T | p.R174C | Breast and/or ovarian cancer | [Théry JC](http://www.ncbi.nlm.nih.gov/pubmed/?term=Th%C3%A9ry%20JC%5BAuthor%5D&cauthor=true&cauthor_uid=21673748),et al 27 |
|  | exon10 | c.1804G>A | p.G602R | Breast and/or ovarian cancer | Michils,et al 28 |
|  | exon11 | c.6235G>T | p.V2079L | Breast and/or ovarian cancer | Lecarpentier,et al 29 |
|  | exon18 | c.8219T>A | p.L2740X | Breast and/or ovarian cancer | Popp,et al 30 |
|  | exon17 | c.7863T>A | p.Y2621X | Breast and/or ovarian cancer | Esteban Cardeñosa E,et al 31 |
| CDH1 | exon12 | c.1901C>T | p.A634V | Gastric cancer | Suriano,et al 32 |
| EPCAM | - | - | - | - | - |
| MLH1 | exon13 | c.1459C>T | p.R487X | Colorectal cancer | Fidalgo,et al 33 |
|  | exon9 | c.704A>T | p.D235V | Colorectal cancer | Hu,et al 34 |
|  | exon4 | c.318C>G | p.S106R | Colorectal cancer | Auclair,et al 35 |
|  | exon10 | c.884+2T>A | Splice defect | Colorectal cancer | Mueller-Koch,et al 36 |
|  | exon14 | c.1614G>A | p.W538X | Colorectal cancer | Yuan,et al 37 |
| MSH2 | exon6 | c.943-1G>C | Splice defect | Colorectal cancer | Mangold,et al 38 |
| MSH6 | exon4 | c.2983G>A | p.E995K | Colorectal cancer | Vahteristo,et al 39 |
|  | exon3 | c.604C>A | p.P202T | Colorectal cancer | [Giráldez MD](http://www.ncbi.nlm.nih.gov/pubmed/?term=Gir%C3%A1ldez%20MD%5BAuthor%5D&cauthor=true&cauthor_uid=20924129),et al 40 |
|  | exon5 | c.3425C>A | p.T1142K | Colorectal cancer | [Pérez-Cabornero L](http://www.ncbi.nlm.nih.gov/pubmed/?term=P%C3%A9rez-Cabornero%20L%5BAuthor%5D&cauthor=true&cauthor_uid=23523604),et al 41 |
|  | exon5 | c.3284G>A | p.R1095H | Colorectal cancer | Kariola,et al 42 |
| MUTYH | exon3 | c.271G>A | p.D91N | Colorectal cancer | Nielsen,et al 43 |
| PMS2 | - | - | - | - | - |
| PTEN | exon8 | c.821G>T | p.W274L | Developmental delay | McBride,et al 44 |
|  | Exon7 | c.697C>T | p.R233X | Cowden disease | Liaw,et al 45 |
| SMAD4 | exon9 | c.988G>T | p.E330X | Juvenile polyposis | Gallione,et al 46 |
|  | exon9 | c.1082G>A | p.R361H | Juvenile polyposis coli | Kim,et al 47 |
| SKT11 | exon4 | c.580G>A | p.D194N | Peutz-Jeghers syndrome | Westerman,et al 48 |
|  | exon8 | c.971C>T | p.P324L | Peutz-Jeghers syndrome | Yoon,et al 49 |
|  | exon3 | c.427G>A | p.V143M | Peutz-Jeghers syndrome | Lim,et al 50 |

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