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| **Supplementary Table 1 Multigene Panel Gene List** | | | | | | |
| **Gene Symbol** | **Gene Name** | **Cancer syndrome(s)** | **Major associated tumour types** | **Mechanism of action of CPG mutations** | **Mode of inheritance** | **Reference (PubMed ID)** |
| ***ABCB11*** | ATP-binding cassette, sub-family B (MDR/TAP), member 11 | Progressive familial intrahepatic cholestasis | Hepatocellular carcinoma Cholangiocarcinoma | loss-of-function | autosomal recessive | 9806540 |
| ***AIP*** | aryl hydrocarbon receptor-interacting protein | Pituitary adenoma | Pituitary adenoma 1, multiple types, Pituitary adenoma predisposition | loss-of-function | autosomal recessive | 16728643 17244780 |
| ***ALK*** | anaplastic lymphoma receptor tyrosine kinase | Neuroblastoma | Neuroblastoma | gain-of-function | autosomal dominant | 18724359 |
| ***ANKRD26*** | ankyrin repeat domain 26 |  | Myeloid malignancies |  | autosomal dominant | 24030261 24628296  28600339 |
| ***APC*** | adenomatous polyposis coli | Familial adenomatous polyposis (FAP) | Colorectal cancer  Hepatoblastoma Desmoid tumour | loss-of-function | autosomal dominant | 1651174 1651562 1651563 1678319 |
| ***ATM*** | ataxia-telangiectasia mutated | Ataxia-Telangiectasia (biallelic mutations) | Biallelic mutations: Lymphoid haematological malignancy (leukaemia, lymphoma)  Monoallelic mutations:  Breast cancer | loss-of-function | autosomal recessive autosomal dominant | 7792600 |
| ***AXIN2*** | axin 2 | oligodentia-colorectal cancer syndrome | Colorectal cancer | loss-of-function | autosomal dominant | 15042511 16110024 27696107 |
| ***BAP1*** | BRCA1 associated protein-1 (ubiquitin carboxy-terminal hydrolase) | Tumour predisposition syndrome | Melanoma (cutaneous, uveal) Mesothelioma Meningioma Lung cancer (adenocarcinoma) | loss-of-function | autosomal dominant | 21874003 |
| ***BARD1*** | BRCA1 associated RING domain 1 | Breast cancer | Breast cancer, Ovarian cancer, Pancreatic cancer and endometrial cancer, colon cancer | loss-of-function | autosomal dominant | 15342711 18481171 20077502 23334666 26010302 26483394 26720728 |
| ***BLM*** | Bloom syndrome, RecQ helicase-like | Bloom syndrome | Lymphoma and ALL haematological malignancy Myeloid haematological malignancy Squamous cell carcinoma, SCC gastric, colorectal cancers | loss-of-function | autosomal recessive | 7585968 |
| ***BMPR1A*** | bone morphogenetic protein receptor, type IA | Juvenile polyposis syndrome | Colorectal cancer, gastric cancer, hamartoma | loss-of-function | autosomal dominant | 11381269 |
| ***BRCA1*** | breast cancer 1, early onset | Hereditary breast-ovarian cancer | Breast cancer Ovarian cancer | loss-of-function | autosomal dominant | 7545954 |

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| --- | --- | --- | --- | --- | --- | --- |
| **Gene Symbol** | **Gene Name** | **Cancer syndrome(s)** | **Major associated tumour types** | **Mechanism of action of CPG mutations** | **Mode of inheritance** | **Reference (PubMed ID)** |
| ***BRCA2*** | breast cancer 2, early onset | Hereditary breast-ovarian cancer Fanconi anaemia (D1) (biallelic mutations) | Biallelic mutations: Myeloid haematological malignancy ( Medulloblastoma Wilms tumour Monoallelic mutations: Breast cancer Ovarian cancer Prostate cancer Pancreas cancer | loss-of-function | autosomal recessive autosomal dominant | 8524414 |
| ***BRIP1*** | BRCA1 interacting protein C-terminal helicase 1 | Fanconi anaemia (J) (biallelic mutations) | Biallelic mutations: Myeloid haematological malignancy  Squamous cell carcinoma (head and neck, oesophagus, genital tract) Monoallelic mutations: Breast cancer Ovarian cancer | loss-of-function | autosomal recessive autosomal dominant | 16153896 16116424 16116423 |
| ***BUB1B*** | budding uninhibited by benzimidazoles 1 homolog beta (yeast) | Mosaic variegated aneuploidy Syndrome | Wilms Tumor  Rhabdomyosarcoma Myeloid haematological malignancy | loss-of-function | autosomal recessive | 15475955 |
| ***CASR*** | calcium-sensing receptor |  | Colorectal cancer, breast cancer and prostate cancer | loss-of-function, a gain of function | autosomal dominant | 26929638 23555732 |
| ***CBL*** | Cbl proto-oncogene, E3 ubiquitin-protein ligase | Noonan syndrome | JMML | loss-of-function | autosomal dominant | 20694012 |
| ***CDC73*** | cell division cycle 73, Paf1/RNA polymerase II complex component, homolog (S. cerevisiae) | Hyperparathyroidism-jaw tumour syndrome | Parathyroid cancer Ossifying fibroma (bone) | loss-of-function | autosomal dominant | 12434154 |
| ***CDH1*** | cadherin 1, type 1, E-cadherin (epithelial) | Hereditary diffuse gastric cancer | Breast cancer (lobular) Gastric cancer (diffuse) | loss-of-function | autosomal dominant | 9537325 |
| ***CDK4*** | cyclin-dependent kinase 4 | Melanoma | Melanoma | gain-of-function | autosomal dominant | 8528263 |
| ***CDKN1B*** | cyclin-dependent kinase inhibitor 1B (p27, Kip1) | Multiple endocrine neoplasias, type IV | Thyroid cancer,  Pituitary adenoma | loss-of-function | autosomal recessive autosomal dominant | 17030811 |
| ***CDKN1C*** | cyclin-dependent kinase inhibitor 1C |  | Wilms tumour and hepatoblastoma | loss-of-function | autosomal dominant | 27419809 |
| ***CDKN2A*** | cyclin-dependent kinase inhibitor 2A | Melanoma and neural system tumour syndrome  Melanoma-pancreatic cancer syndrome | Melanoma [p16 and p14ARF] Pancreas cancer [p16 ] Astrocytoma [p14ARF ] | loss-of-function | autosomal dominant | 7987387 7987388 |
| ***CEBPA*** | CCAAT/enhancer-binding protein (C/EBP), alpha | Leukaemia, acute myeloid | Myeloid haematological malignancy | loss-of-function | autosomal dominant | 15575056 |
| ***CFTR*** | CF transmembrane conductance regulator |  | Colorectal cancer, pancreatic cancer, | loss-of-function | autosomal dominant | 14576497 26751771 |
| ***CHEK2*** | checkpoint kinase 2 | Breast cancer, Prostate cancer | Breast cancer, Prostate cancer and colorectal cancer | loss-of-function | autosomal dominant | 11967536 12094328 |
| ***COL7A1*** | collagen, type VII, alpha 1 | Epidermolysis bullosa | Squamous cell carcinoma (skin) | loss-of-function | autosomal recessive autosomal dominant | 8513326 |
| ***CTRC*** | chymotrypsin C |  | Pancreatic cancer | loss-of-function | autosomal dominant | 24600409 30134356 |

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| **Gene Symbol** | **Gene Name** | **Cancer syndrome(s)** | **Major associated tumour types** | **Mechanism of action of CPG mutations** | **Mode of inheritance** | **Reference (PubMed ID)** |
| ***CYLD*** | cylindromatosis (turban tumour syndrome) | Brooke-Spiegler syndrome | Cylindroma spiroadenocarcinoma Basal cell carcinoma | loss-of-function | autosomal dominant | 10835629 |
| ***DDB2*** | damage-specific DNA binding protein 2, 48kDa | Xeroderma Pigmentosum (E) | Basal cell carcinoma Squamous cell carcinoma Melanoma | loss-of-function | autosomal dominant | 8798680 |
| ***DICER1*** | dicer 1, ribonuclease type III | DICER1 syndrome | Pleuropulmonary blastoma Cystic nephroma  Ovarian sex cord tumour | loss-of-function | autosomal dominant | 19556464 |
| ***DIS3L2*** | DIS3 mitotic control homolog (S. cerevisiae)-like 2 | Perlman syndrome | Wilms tumour | loss-of-function | autosomal recessive | 22306653 |
| ***DKC1*** | dyskeratosis congenita 1, dyskerin | Dyskeratosis congenita | acute myeloid leukaemia Squamous cell carcinoma (head + neck, anorectal) | loss-of-function | X-linked recessive | 9590285 |
| ***DOCK8*** | dedicator of cytokinesis 8 | HyperIgE syndrome | Squamous cell carcinoma Lymphoma | loss-of-function | autosomal recessive | 19776401 |
| ***EGFR*** | epidermal growth factor receptor |  | Non-small cell lung cancer | gain-of-function | autosomal dominant | 16258541 |
| ***ELANE*** | elastase, neutrophil expressed | Severe congenital neutropenia | Leukaemia | loss-of-function | autosomal dominant | 11001877 |
| ***EPCAM*** | epithelial cell adhesion molecule | Colorectal cancer, hereditary nonpolyposis, Lynch syndrome | Colorectal cancer, Endometrial cancer, Prostate cancer, Gastric cancer, and Ovarian cancer | loss-of-function | autosomal dominant |  |
| ***ERCC2*** | excision repair cross-complementing rodent repair deficiency, complementation group 2 | Xeroderma pigmentosum (D) | Basal cell carcinoma Squamous cell carcinoma Melanoma | loss-of-function | autosomal recessive | 7849702 |
| ***ERCC3*** | excision repair cross-complementing rodent repair deficiency, complementation group 3 | Xeroderma pigmentosum (B) | Basal cell carcinoma Squamous cell carcinoma Melanoma | loss-of-function | autosomal recessive | 2167179 |
| ***ERCC4*** | excision repair cross-complementing rodent repair deficiency, complementation group 4 | Xeroderma pigmentosum (F) Fanconi anaemia (Q) | Basal cell carcinoma Squamous cell carcinoma Melanoma | loss-of-function | autosomal recessive | 8797827 |
| ***ERCC5*** | excision repair cross-complementing rodent repair deficiency, complementation group 5 | Xeroderma pigmentosum (G) | Basal cell carcinoma Squamous cell carcinoma Melanoma | loss-of-function | autosomal recessive | 7951246 |
| ***EXT1*** | exostosin 1 | Chondrosarcoma | Chondrosarcoma | loss-of-function | autosomal dominant | 7550340 |
| ***EXT2*** | exostosin 2 |  | Chondrosarcoma | loss-of-function | autosomal dominant | 8782816 |
| ***FAH*** | fumarylacetoacetate hydrolase (fumarylacetoacetase) | Tyrosinemia | Hepatocellular carcinoma | loss-of-function | autosomal recessive | 8318997 |
| ***FANCA*** | Fanconi anemia, complementation group A | Fanconi anaemia (A) | Myeloid haematological malignancy Squamous cell carcinoma (head and neck, oesophagus, genital tract) | loss-of-function | autosomal recessive | 8896564 8896563 |
| ***FANCC*** | Fanconi anemia, complementation group C | Fanconi anaemia (C) | Myeloid haematological malignancy Squamous cell carcinoma (head and neck, oesophagus, genital tract) | loss-of-function | autosomal recessive | 1574115 |
| ***FANCG*** | Fanconi anemia, complementation group G | Fanconi anaemia (G) | Myeloid haematological malignancy Squamous cell carcinoma (head and neck, oesophagus, genital tract) | loss-of-function | autosomal recessive | 9806548 |
| ***FH*** | fumarate hydratase | Hereditary leiomyomatosis and renal cell cancer (HLRCC) | Renal cell cancer Leiomyosarcoma (uterus) | loss-of-function | autosomal recessive autosomal dominant | 11865300 |

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| **Gene Symbol** | **Gene Name** | **Cancer syndrome(s)** | **Major associated tumour types** | **Mechanism of action of CPG mutations** | **Mode of inheritance** | **Reference (PubMed ID)** |
| ***FLCN*** | folliculin | Birt-Hogg-Dube syndrome | Renal cell cancer Oncocytoma | loss-of-function | autosomal dominant | 12204536 |
| *GALNT12* | polypeptide N-acetylgalactosaminyltransferase 12 | Colorectal cancer | Colorectal cancer | loss-of-function | autosomal dominant | 19617566 |
| ***GATA2*** | GATA binding protein 2 | Emberger MonoMAC syndrome | Myeloid haematological malignancy | loss-of-function | autosomal dominant | 21892158 21892162 21765025 21670465 |
| ***GBA*** | glucosidase, beta, acid | Gauchers type 1 | Myeloma Lymphoma Hepatocellular carcinoma | loss-of-function | autosomal recessive | 2880291 |
| ***GJB2*** | gap junction protein, beta 2, 26kDa | Keratosis-ichthyosis-deafness syndrome (KID) | Squamous cell carcinoma | loss-of-function | autosomal dominant | 11912510 |
| ***GPC3*** | glypican 3 | Simpson-Golabi-Behmel syndrome | Wilms tumour Hepatoblastoma, hepatocellular carcinoma Neuroblastoma Gonadoblastoma | loss-of-function | X-linked recessive | 8589713 |
| ***GREM1*** | gremlin 1, DAN family BMP antagonist | Hereditary mixed polyposis syndrome (AD) | Hamartomatous polyps, Adenomatous polyps Colorectal cancer | gain-of-function | autosomal dominant | 22561515 26493165 |
| ***HFE*** | hemochromatosis | Haemochromatosis | Hepatocellular carcinoma Cholangiocarcinoma | loss-of-function | autosomal recessive | 8696333 |
| ***HMBS*** | hydroxymethylbilane synthase | Porphyria (AI) | hepatocellular carcinoma | loss-of-function | autosomal dominant | 2563167 |
| ***HOXB13*** | homeobox B13 | Prostate cancer | Prostate cancer | loss-of-function gain-of-function | autosomal dominant | 22236224 |
| ***HRAS*** | v-Ha-ras Harvey rat sarcoma viral oncogene homolog | Costello syndrome | Rhabdomyosarcoma Neuroblastoma Transitional cell carcinoma (bladder) | gain-of-function | autosomal dominant | 16170316 |
| ***ITK*** | IL2-inducible T-cell kinase | Lymphoproliferative syndrome 1 | Hodgkins lymphoma | loss-of-function | autosomal recessive | 19425169 |
| ***KIT*** | v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog | Gastrointestinal stromal tumour, familial | Gastro-Intestinal Stromal Tumor | gain-of-function | autosomal dominant | 9697690 |
| ***MAX*** | MYC associated factor X | Familial paraganglioma-pheochromocytoma syndrome | Paraganglioma Pheochromocytoma | loss-of-function | autosomal dominant | 21685915 |
| ***MEN1*** | multiple endocrine neoplasia I | Multiple endocrine neoplasia Type 1 | Parathyroid, pituitary adenoma  Neuroendocrine tumour Carcinoid tumour Adrenocortical carcinoma | loss-of-function | autosomal dominant | 9103196 |
| ***MET*** | met proto-oncogene (hepatocyte growth factor receptor) | Renal cell carcinoma, papillary, 1, familial and somatic | Renal cell cancer (papillary carcinoma) Osteofibrous dysplasia | gain-of-function | autosomal dominant | 9140397 |
| ***MLH1*** | mutL homolog 1, colon cancer, nonpolyposis type 2 (E. coli) | MMR deficiency syndrome (biallelic mutations) Lynch syndrome / Hereditary Non-Polyposis Colon Cancer (monoallelic mutations) | Biallelic mutations: Brain tumours Haematological malignancy Embryonal tumours Monoallelic mutations: Colorectal cancer Endometrial cancer Ovarian cancer | loss-of-function | autosomal recessive autosomal dominant | 8128251 8145827 |

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| **Gene Symbol** | **Gene Name** | **Cancer syndrome(s)** | **Major associated tumour types** | **Mechanism of action of CPG mutations** | **Mode of inheritance** | **Reference (PubMed ID)** |
| ***MSH2*** | mutS homolog 2, colon cancer, nonpolyposis type 1 (E. coli) | MMR deficiency syndrome (biallelic mutations) Lynch syndrome / Hereditary Non-Polyposis Colon Cancer (monoallelic mutations) | Biallelic mutations: Brain tumours Haematological malignancy Embryonal tumours Monoallelic mutations: Colorectal cancer Endometrial cancer Ovarian cancer Sebaceous adenoma, carcinoma, epithelioma | loss-of-function | autosomal recessive autosomal dominant | 8252616 8261515 |
| ***MSH3*** | mutS homolog 3 | Familial adenomatous polyposis | Colorectal cancer | loss-of-function | autosomal recessive | 27476653 |
| ***MSH6*** | mutS homolog 6 (E. coli) | MMR deficiency syndrome (biallelic mutations) Lynch syndrome / Hereditary Non-Polyposis Colon Cancer (monoallelic mutations) | Biallelic mutations: Brain tumours Haematological malignancy Embryonal tumours Monoallelic mutations: Colorectal cancer Endometrial cancer Ovarian cancer | loss-of-function | autosomal recessive autosomal dominant | 9354786 |
| ***MTAP*** | methylthioadenosine phosphorylase | Diaphyseal medullary stenosis with malignant fibrous histiocytoma (DMS-MFH) | malignant fibrous histiocytoma (sarcoma) | loss-of-function | autosomal dominant | 22464254 |
| ***MUTYH*** | mutY homolog (E. coli) | Adenomas, multiple colorectal | Colorectal cancer | loss-of-function | autosomal recessive | 11818965 |
| ***NBN*** | nibrin | Nijmegen breakage syndrome | Lymphoma  Medulloblastoma Glioma Rhabdomyosarcoma | loss-of-function | autosomal recessive | 9590180 9620777 |
| ***NF1*** | neurofibromin 1 | Neurofibromatosis type 1 | Glioma Malignant peripheral nerve sheath tumour | loss-of-function | autosomal dominant | 2134734 1694727 |
| ***NF2*** | neurofibromin 2 (merlin) | Neurofibromatosis type 2 | Vestibular schwannoma Meningioma Ependymoma | loss-of-function | autosomal dominant | 8453669 8379998 |
| ***NTHL1*** | nth like DNA glycosylase 1 | Familial adenomatous polyposis 3 | Colorectal cancer Endometrial cancer | loss-of-function | autosomal recessive | 25938944 27720914 27713038 30248171 |
| ***PALB2*** | partner and localizer of BRCA2 | Fanconi anaemia (N) (biallelic mutations) | Biallelic mutations: Myeloid haematological malignancy  Medulloblastoma Neuroblastoma Wilms tumour Monoallelic mutations: Breast cancer Pancreas cancer | loss-of-function | autosomal recessive autosomal dominant | 17200671 17200672 17200668 17287723 |
| ***PALLD*** | palladin, cytoskeletal associated protein | Pancreatic cancer | Pancreatic cancer | gain-of-function | autosomal dominant | 17194196 17415588 19336541 |
| ***PDGFRA*** | platelet-derived growth factor receptor, alpha polypeptide |  | Gastro-Intestinal Stromal Tumor | gain-of-function | autosomal dominant | 14699510 |
| ***PHOX2B*** | paired-like homeobox 2b |  | Neuroblastoma | loss-of-function | autosomal dominant | 12640453 |
| **Gene Symbol** | **Gene Name** | **Cancer syndrome(s)** | **Major associated tumour types** | **Mechanism of action of CPG mutations** | **Mode of inheritance** | **Reference (PubMed ID)** |
| ***PMS2*** | PMS2 postmeiotic segregation increased 2 (S. cerevisiae) | MMR deficiency syndrome (biallelic mutations) Lynch syndrome / Hereditary Non-Polyposis Colon Cancer (monoallelic mutations) | Biallelic mutations: Brain tumours  Haematological malignancy Supratentorial primitive neuroectodermal tumors Monoallelic mutations: Colorectal cancer Endometrial cancer Ovarian cancer | loss-of-function | autosomal recessive autosomal dominant | 8072530 |
| ***POLD1*** | polymerase (DNA directed), delta 1, catalytic subunit | PPAP (polymerase proofreading associated polyposis) | Colorectal cancer Endometrial cancer | loss-of-function | autosomal dominant | 23263490 |
| ***POLE*** | polymerase (DNA directed), epsilon, catalytic subunit | PPAP (polymerase proofreading associated polyposis) | Colorectal cancer | loss-of-function | autosomal recessive autosomal dominant | 23263490 |
| ***POLH*** | polymerase (DNA directed), eta | Xeroderma pigmentosa V | Squamous cell cancer (skin) | loss-of-function | autosomal recessive | 10385124 |
| ***POT1*** | protection of telomeres 1 | Glioma, Melanoma | Cutaneous malignant melanoma thyroid cancer Li-Fraumeni-like syndrome | loss-of-function | autosomal recessive | 28389767 24686846 24686849 25482530 |
| ***PRKAR1A*** | protein kinase, cAMP-dependent, regulatory, type I, alpha | Carney complex | Myxoma (cardiac/cutaneous/breast) Thyroid cancer Sex cord-stromal tumor | loss-of-function | autosomal dominant | 10973256 |
| ***PRSS1*** | protease, serine, 1 (trypsin 1) |  | Pancreatic cancer | loss-of-function | autosomal dominant | 8841182 |
| ***PTCH1*** | patched 1 | Nevoid basal cell carcinoma syndrome Gorlin Syndrome | Basal cell carcinoma Medulloblastoma | loss-of-function | autosomal dominant | 8658145 8681379 |
| ***PTEN*** | phosphatase and tensin homolog | Cowden Syndrome PTEN hamartoma tumor syndrome | Breast cancer Thyroid cancer Endometrial cancer | loss-of-function | autosomal dominant | 9140396 |
| ***PTPN11*** | protein tyrosine phosphatase, non-receptor type 11 | Noonan syndrome | JMML neuroblastoma | gain-of-function | autosomal dominant | 11704759 12717436 |
| ***RAD51C*** | RAD51 homolog C (S. cerevisiae) | Fanconi anaemia (O) (biallelic mutations) | Monoallelic mutations:  Ovarian cancer | loss-of-function | autosomal recessive autosomal dominant | 20400964 |
| ***RAD51D*** | RAD51 homolog D (S. cerevisiae) |  | Ovarian cancer | loss-of-function | autosomal dominant | 21822267 |
| ***RB1*** | retinoblastoma 1 | retinoblastoma | Retinoblastoma Pinealoma Sarcoma Melanoma | loss-of-function | autosomal dominant | 2885916 |
| ***RECQL4*** | RecQ protein-like 4 | Rothmund-Thompson syndrome | Osteosarcoma Basal cell carcinoma Squamous cell carcinoma | loss-of-function | autosomal recessive | 10319867 |
| ***RET*** | ret proto-oncogene | Multiple endocrine neoplasia 2A/2B  Familial medullary thyroid carcinoma | Medullary thyroid cancer Pheochromocytoma | gain-of-function | autosomal dominant | 8099202 |
| ***RHBDF2*** | rhomboid 5 homolog 2 (Drosophila) |  | Oesophagal cancer | gain-of-function | autosomal dominant | 22265016 |
| ***RMRP*** | The RNA component of mitochondrial RNA processing endoribonuclease | Cartilage-hair hypoplasia syndrome | Non-Hodgkin lymphoma Squamous carcinoma (bcc) Leukaemia | loss-of-function | autosomal recessive | 11207361 |
| **Gene Symbol** | **Gene Name** | **Cancer syndrome(s)** | **Major associated tumour types** | **Mechanism of action of CPG mutations** | **Mode of inheritance** | **Reference (PubMed ID)** |
| ***RNF43*** | ring finger protein 43 | Sessile serrated polyposis cancer syndrome | Colorectal cancer | loss-of-function | autosomal dominant | 24512911 22895187 27081527 |
| *RPS20* | ribosomal protein S20 |  | hereditary nonpolyposis CRC | loss-of-function | autosomal dominant | 24941021 27713038 |
| ***RUNX1*** | runt-related transcription factor 1 |  | Myeloid haematological malignancy (leukaemia) | loss-of-function | autosomal dominant | 10508512 |
| ***SBDS*** | Shwachman-Bodian-Diamond syndrome | Schwachman-Diamond syndrome | Myeloid haematological malignancy | loss-of-function | autosomal recessive | 12496757 |
| ***SDHA*** | succinate dehydrogenase complex, subunit A, flavoprotein (Fp) | Carney-Stratakis syndrome | Paraganglioma Pheochromocytoma A gastrointestinal stromal tumour (GIST) | loss-of-function | autosomal recessive autosomal dominant | 20484225 |
| ***SDHAF2*** | succinate dehydrogenase complex assembly factor 2 | Familial paraganglioma-pheochromocytoma syndrome | Paraganglioma Pheochromocytoma | loss-of-function | autosomal dominant | 19628817 |
| ***SDHB*** | succinate dehydrogenase complex, subunit B, iron sulfur (Ip) | Familial paraganglioma-pheochromocytoma syndrome | Paraganglioma Pheochromocytoma Renal cell cancer | loss-of-function | autosomal dominant | 11404820 |
| ***SDHC*** | succinate dehydrogenase complex, subunit C, integral membrane protein, 15kDa | Familial paraganglioma-pheochromocytoma syndrome | Paraganglioma Pheochromocytoma Gastrointestinal stromal tumour (GIST) | loss-of-function | autosomal dominant | 11062460 |
| ***SDHD*** | succinate dehydrogenase complex, subunit D, integral membrane protein | Familial paraganglioma-pheochromocytoma syndrome | Paraganglioma Pheochromocytoma Gastrointestinal stromal tumour (GIST) | loss-of-function | autosomal dominant | 10657297 |
| ***SERPINA1*** | serpin peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 1 | Alpha1 antitrypsin deficiency | Hepatocellular carcinoma | loss-of-function | autosomal recessive | 3485248 7045697 |
| ***SH2D1A*** | SH2 domain-containing 1A | Lymphoproliferative disease | Lymphoma | loss-of-function | X-linked recessive | 9771704 |
| ***SLC25A13*** | solute carrier family 25 (aspartate/glutamate carrier), member 13 | Citrullinaemia | Hepatocellular carcinoma | loss-of-function | autosomal recessive | 10369257 |
| ***SMAD4*** | SMAD family member 4 | Juvenile polyposis syndrome | Colorectal cancer | loss-of-function | autosomal dominant | 9582123 |
| ***SMARCB1*** | SWI/SNF related, matrix associated, actin-dependent regulator of chromatin, subfamily b, member 1 | Rhabdoid predisposition syndrome | Rhabdoid tumour (renal, extra-renal) Central primitive neuroectodermal tumour | loss-of-function | autosomal dominant | 10521299 |
| ***SMARCE1*** | SWI/SNF related, matrix associated, actin-dependent regulator of chromatin, subfamily e, member 1 |  | Meningioma | loss-of-function | autosomal dominant | 23377182 |
| ***SOS1*** | son of sevenless homolog 1 (Drosophila) | Noonan syndrome | Rhabdomyosarcoma | gain-of-function | autosomal dominant | 17143285 |
| ***SPINK1*** | serine peptidase inhibitor Kazal type 1 |  | Pancreatic cancer Colorectal cancer Breast cancer | gain-of-function | autosomal recessive autosomal dominant | 26656134 |
| ***SRY*** | sex-determining region Y |  | Gonadoblastoma | loss-of-function | Y-linked | 2247149 2247151 |
| ***STAT3*** | signal transducer and activator of transcription 3 (acute-phase response factor) | Hyper-immunoglobulin E syndrome | Lymphoma | loss-of-function | autosomal dominant | 17676033 |
| ***STK11*** | serine/threonine kinase 11 | Peutz-Jeghers syndrome | Colorectal cancer Gastric cancer Breast cancer Sex cord-stromal tumor | loss-of-function | autosomal dominant | 9425897 9428765 |
| ***SUFU*** | suppressor of fused homolog (Drosophila) |  | Medulloblastoma, meningioma | loss-of-function | autosomal dominant | 12068298 |
| **Gene Symbol** | **Gene Name** | **Cancer syndrome(s)** | **Major associated tumour types** | **Mechanism of action of CPG mutations** | **Mode of inheritance** | **Reference (PubMed ID)** |
| ***TERT*** | telomerase reverse transcriptase | Dyskeratosis congenita | acute myeloid leukaemia  Squamous cell carcinoma (head + neck, anorectal)  Melanoma | loss-of-function | autosomal recessive autosomal dominant | 16247010 |
| ***TGFBR1*** | transforming growth factor, beta receptor 1 | Multiple self-healing squamous epithelioma (MSSE) Ferguson-Smith syndrome | Squamous cell carcinoma (skin) | loss-of-function | autosomal dominant | 21358634 |
| ***TMEM127*** | transmembrane protein 127 |  | Pheochromocytoma | loss-of-function | autosomal dominant | 20154675 |
| ***TNFRSF6 (FAS)*** | transforming growth factor, beta receptor 1 | Autoimmune lymphoproliferative syndrome | Lymphoma | loss-of-function | autosomal dominant | 7540117 |
| ***TP53*** | tumour protein p53 | Li-Fraumeni syndrome | Breast cancer Sarcoma Adrenocortical carcinoma Astrocytoma | loss-of-function | autosomal dominant | 1978757 |
| ***TRIM37*** | tripartite motif containing 37 | Mulibrey-nanism | Wilms tumour | loss-of-function | autosomal recessive | 10888877 |
| ***TSC2*** | tuberous sclerosis 2 | Tuberous sclerosis 2 | Renal cell cancer, angiomyolipoma Subependymal giant cell astrocytoma Rhabdomyoma (cardiac) | loss-of-function | autosomal dominant | 8269512 |
| ***UROD*** | uroporphyrinogen decarboxylase | Porphyria (cutanea tarda) | hepatocellular carcinoma | loss-of-function | autosomal recessive autosomal dominant | 3775362 |
| ***VHL*** | von Hippel-Lindau tumour suppressor, E3 ubiquitin protein ligase | Von Hippel-Lindau syndrome | Renal cell cancer Pheochromocytoma Neuroendocrine tumour (pancreas) Hemangioblastoma (central nervous system, retina) | loss-of-function | autosomal dominant | 8493574 |
| ***WAS*** | Wiskott-Aldrich syndrome | Wiskott-Aldrich syndrome WAS-related syndrome | Lymphoma | loss-of-function | X-linked recessive | 8069912 |
| ***WRN*** | Werner syndrome, RecQ helicase-like | Werner syndrome | Sarcoma Melanoma Thyroid cancer | loss-of-function | autosomal recessive | 8602509 |
| ***WT1*** | Wilms tumour 1 | WAGR syndrome Denys-Drash syndrome Frasier syndrome | Wilms tumour Gonadoblastoma | loss-of-function | autosomal dominant | 1673293 |
| ***XPA*** | xeroderma pigmentosum, complementation group A | Xeroderma pigmentosum (A) | Basal cell carcinoma Squamous cell carcinoma Melanoma | loss-of-function | autosomal recessive | 2234061 |
| ***XPC*** | xeroderma pigmentosum, complementation group C | Xeroderma pigmentosum (C) | Basal cell carcinoma Squamous cell carcinoma Melanoma | loss-of-function | autosomal recessive | 8298653 |

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| **Supplementary Table 2.** Association of characteristics with patients with proximal colon cancer vs distal colon cancer. | | | |
| **Features (No. of the patient)** | **% Proximal colon cancer** | **% Distal colon cancer** | **P-Value** |
| Male (62) | 14.5 | 85.5 | 0.003 |
| Female (65) | 38.5 | 61.5 |
| FDR with CRC (18) | 38.9 | 61.1 | 0.3 |
| NO FDR with CRC (109) | 24.8 | 75.2 |
| FDR with BC (16) | 50 | 50 | 0.03 |
| NO FDR with BC (111) | 23.4 | 76.6 |
| FDR with T2D (43) | 20.9 | 79.1 | 0.4 |
| No FDR with T2D (84) | 29.8 | 70.2 |
| With *KRAS* variants (30) | 36.7 | 63.3 | 0.3 |
| Without *KRAS* variants (46) | 21.7 | 78.3 |
| With *BRAF* variants (9) | 44.4 | 55.6 | 0.4 |
| Without *BRAF* variants (68) | 26.5 | 73.5 |
| Ever smoked (64) | 18.8 | 81.2 | 0.1 |
| Never smoked (60) | 35 | 65 |
| Current smoker (18) | 5.6 | 94.4 | 0.09 |
| Non-smoker (106) | 30.2 | 69.8 |

BC: Breast cancer; CRC: Colorectal cancer; FDR: First-degree relative; and T2D: Type 2 diabetes

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| **Supplementary Table 3.** Germline variants in young adults with CRC with evidence of pathogenicity according to the ACMG guidelines. | | | | | | | | |
| Chr | Start | Ref | Alt | Gene | cDNA | AA Change | ACMG criteria | Classification |
| 1 | 45797348 | G | A | MUTYH | NM\_001048171.1:c.1129C>T | p.(Gln377\*) | PVS1, PS1, PS3, PM2 | Pathogenic |
| 1 | 45799124 | C | T | MUTYH | NM\_001048171.1:c.267G>A | p.(Trp89\*) | PVS1, PM2 | Likely-pathogenic |
| 1 | 45797228 | C | T | MUTYH | NM\_001048171.1:c.1145G>A | p.(Gly382Asp) | PS1, PS3, PP3 | Pathogenic |
| 1 | 45798475 | T | C | MUTYH | NM\_001048171.1:c.494A>G | p.(Tyr165Cys) | PS1, PS3, PP1, PP3 | Pathogenic |
| 2 | 48030691 | CT | C | MSH6 | NM\_000179.2:c.3312del | p.(Phe1104Leufs\*11) | PVS1, PS1, PM2, PP4 | Pathogenic |
| 1 | 45797752 | G | A | MUTYH | NM\_001048171.1:c.898C>T | p.(Gln300\*) | PVS1, PS1, PM2 | Pathogenic |
| 2 | 47703538 | C | T | MSH2 | NM\_000251.2:c.2038C>T | p.(Arg680\*) | PVS1, PS1, PM2, PP4 | Pathogenic |
| 2 | 48033752 | A | AGAAT | MSH6 | c.3964\_3967dup | p.(Phe1323\*) | PVS1, PS1, PM2, PP4 | Pathogenic |
| 9 | 98011506 | TC | T | FANCC | NM\_000136.2:c.67del | p.(Asp23Ilefs\*23) | PVS1, PS1, PM2 | Pathogenic |
| 3 | 37045935 | C | T | MLH1 | NM\_000249.3:c.350C>T | p.(Thr117Met) | PS1, PS3, PM2, PP3, PP4 | Pathogenic |
| 2 | 47641560 | A | T | MSH2 | NM\_000251.2:c.942+3A>T | p.? | PVS1, PS1, PS3, PM2, PP4 | Pathogenic |
| 10 | 88671996 | G | T | BMPR1A | NM\_004329.2:c.531-1G>T | p.? | PVS1, PM2 | Likely-pathogenic |
| 17 | 56448271 | C | T | RNF43 | NM\_017763.5:c.375+1G>A | p.? | PVS1\_M, PM2, PS3, PP4 | Likely-pathogenic |
| 13 | 32914209 | ACT | A | BRCA2 | NM\_000059.3:c.5722\_5723del | p.(Leu1908Argfs\*2) | PVS1, PS1, PS3, PM2 | Pathogenic |
| 13 | 32913365 | GAA | G | BRCA2 | NM\_000059.3:c.4876\_4877del | p.(Asn1626Serfs\*12) | PVS1, PS1, PS3, PM2 | Pathogenic |
| 13 | 32968967 | C | G | BRCA2 | NM\_000059.3:c.9398C>G | p.(Ser3133\*) | PVS1, PM2 | Likely-pathogenic |
| 13 | 32968863 | C | G | BRCA2 | NM\_000059.3:c.9294C>G | p.(Tyr3098\*) | PVS1, PS1, PS3, PM2 | Pathogenic |
| 11 | 108183193 | AAAAGT | A | ATM | NM\_000051:c.5979\_5983del | p.(Ser1993Argfs\*23) | PVS1, PS1, PS4, PM2 | Pathogenic |
| 17 | 59793412 | G | A | BRIP1 | NM\_032043.2:c.2392C>T | p.(Arg798\*) | PVS1, PS1, PS3, PM2 | Pathogenic |
| 17 | 56780641 | T | C | RAD51C | NM\_058216.2:c.656T>C | p.(Leu219Ser) | PS1, PS3, PS4, PM2, PP3 | Pathogenic |
| 19 | 45855589 | G | A | ERCC2 | NM\_000400.3:c.2068C>T | p.(Arg690Trp) | PS1, PM2, PS3, PP3 | Pathogenic |
| 3 | 128204915 | T | G | GATA2 | NM\_001145661.1.c.526A>C | p.(Thr176Pro) | PP1-S, PM2, PM6, PP3 | Likely-pathogenic |
| 13 | 20763620 | A | G | GJB2 | c.101T>C | p.(Met34Thr) | PS1, PS3, PP1-S, PP3 | Pathogenic |
| 16 | 14029554 | C | T | ERCC4 | c.1765C>T | p.(Arg589Trp) | PSI, PM2, PS3, PP3 | Pathogenic |
| 7 | 117180284 | C | T | CFTR | NM\_000492.3:c.1000C>T | p.(Arg334Trp) | PS1, PS3, PP1, PM2, PP3 | Pathogenic |
| 7 | 17149101 | G | T | CFTR | NM\_000492.3:c.178G>T | p.(E60\*) | PVS1, PS1, PS3, PM2 | Pathogenic |

Chr: Chromosome; Ref: Reference allele; Alt: Alternative allele; AA: Amino acid; and ACMG: The American College of Medical Genetics and Genomics.

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| **Supplementary Table 4. Variants of unknown significance in cancer-associated genes detected in young adults with CRC.** | | | | | | | | |
| Patient | Chr | Start | Ref | Alt | *Gene* | Transcript | cDNA | AA Change |
| SAYO 018100 | 9 | 420563 | G | T | *DOCK8* | NM\_001190458.1 | c.3703G>T | p.(Val1235Leu) |
| SAYO\_015028 | 7 | 6038762 | C | T | *PMS2* | NM\_000535.6 | c.682G>A | p.(Gly228Ser) |
| SAYO 014001 | 8 | 90993086 | CAAG | C | *NBN* | NM\_002485.4 | c.353\_355del | p.(Ser118del) |
| 19 | 45856019 | C | G | *ERCC2* | NM\_000400.3 | c.1887G>C | p.(Gln629His) |
| SAYO 015002 | 12 | 1.33E+08 | AG | A | *POLE* | NM\_006231.3 | c.5697del | p.(Phe1900Serfs\*4) |
| SAYO 015007 | 4 | 41747904 | C | T | *PHOX2B* | NM\_003924.3 | c.865G>A | p.(Gly289Ser) |
| SAYO 015014 | 2 | 47707945 | A | G | *MSH2* | NM\_000251.2 | c.2569A>G | p.(Ile857Val) |
| 19 | 1207006 | A | G | *STK11* | NM\_000455.4 | c.94A>G | p.(Thr32Ala) |
| SAYO 015017 | 16 | 89805934 | CGGA | C | *FANCA* | NM\_000135.2 | c.3959\_3961del | p.(Leu1320del) |
| SAYO 016043 | 19 | 11097598 | A | C | *SMARCA4* | NM\_001128844.1 | c.778A>C | p.(Met260Leu) |
| SAYO 017056 | 8 | 30977876 | G | GGTC | *WRN* | NM\_000553.4 | c.2569\_2571dup | p.(Arg857dup) |
| SAYO 018059 | 1 | 17349129 | T | C | *SDHB* | NM\_003000.2 | c.739A>G | p.(Met247Val) |
| SAYO 018060 | 17 | 59926584 | A | G | *BRIP1* | NM\_032043.2 | c.413T>C | p.(Leu138Ser) |
| SAYO 018063 | 8 | 1.46E+08 | C | T | *RECQL4* | NM\_004260.3 | c.3185G>A | p.(Arg1062Gln) |
| 19 | 33792754 | GGGCGGCGGC | G | *CEBPA* | NM\_004364.4 | c.558\_566del | p.(Pro187\_Pro189del) |
| SAYO 018065 | 8 | 31000191 | C | T | *WRN* | NM\_000553.4 | c.3283C>T | p.(Pro1095Ser) |
| SAYO 018070 | 3 | 14187542 | G | A | *XPC* | NM\_001145769.1 | c.2611C>T | p.(Arg871\*) |
| SAYO 018072 | 2 | 48033640 | C | T | *MSH6* | NM\_000179.2 | c.3851C>T | p.(Thr1284Met) |
| 9 | 1.36E+08 | G | A | *TSC1* | NM\_000368.4 | c.1922C>T | p.(Pro641Leu) |
| 14 | 95571450 | C | T | *DICER1* | NM\_001195573.1 | c.3227G>A | p.(Ser1076Asn) |
| SAYO 018074 | 22 | 29095881 | C | T | *CHEK2* | NM\_001005735.1 | c.1082G>A | p.(Arg361His) |
| SAYO 018077 | 17 | 33443879 | G | A | *RAD51D* | NM\_001142571.1 | c.322C>T | p.(Arg108Cys) |
| SAYO 018083 | 9 | 420563 | G | T | *DOCK8* | NM\_001190458.1 | c.3703G>T | p.(Val1235Leu) |
| 9 | 98011565 | T | A | *FANCC* | NM\_000136.2 | c.9A>T | p.(Gln3His) |
| 17 | 41246198 | T | A | *BRCA1* | NM\_007294.3 | c.1350A>T | p.(Lys450Asn) |
| SAYO 018086 | 9 | 420563 | G | T | *DOCK8* | NM\_001190458.1 | c.3703G>T | p.(Val1235Leu) |
| SAYO018087 | 15 | 80450427 | T | C | *FAH* | NM\_000137.2 | c.107T>C | p.(Ile36Thr) |
| SAYO 018088 | 14 | 65543267 | C | T | *MAX* | NM\_001320415.1 | c.221G>A | p.(Gly74Asp) |
| 17 | 59760943 | C | T | *BRIP1* | NM\_032043.2 | c.3464G>A | p.(Gly1155Glu) |
| SAYO 018094 | 2 | 48023078 | C | G | *MSH6* | NM\_000179.2 | c.503C>G | p.(Ala168Gly) |
| 2 | 48026159 | C | T | *MSH6* | NM\_000179.2 | c.1037C>T | p.(Ser346Phe) |
| SAYO 018096 | 13 | 32929030 | C | A | *BRCA2* | NM\_000059.3 | c.7040C>A | p.(Pro2347Gln) |
| SAYO 019106 | 2 | 47630477 | C | G | *MSH2* | NM\_000251.2 | c.147C>G | p.(Asp49Glu) |
| 16 | 2097827 | C | G | *NTHL1* | NM\_002528.6 | c.22G>C | p.(Gly8Arg) |
| SAYO 019109 | 15 | 91347479 | T | C | *BLM* | NM\_000057.3 | c.3641T>C | p.(Met1214Thr) |
| SAYO 019112 | 12 | 1.33E+08 | C | T | *POLE* | NM\_006231.3 | c.2706+5G>A | p.? |
| 19 | 11170550 | C | G | *SMARCA4* | NM\_001128844.1 | c.4757C>G | p.(Ser1586Cys) |
| SAYO019192 | 12 | 11254185 | GGA | G | *POLE* | NM\_006231.3 | c.5697del | p.(Phe1900Serfs\*4) |
| SAYO 019116 | 9 | 98011431 | A | G | *FANCC* | NM\_000136.2 | c.143T>C | p.(Met48Thr) |
| 16 | 89836642 | C | T | *FANCA* | NM\_000135.2 | c.2248G>A | p.(Val750Met) |
| SAYO 019118 | 4 | 55575650 | C | G | *KIT* | NM\_000222.2 | c.1176C>G | p.(Phe392Leu) |
| 15 | 40492521 | C | T | *BUB1B* | NM\_001211.5 | c.1478C>T | p.(Thr493Ile) |
| SAYO019122 | 11 | 1.08E+08 | A | G | *ATM* | NM\_000051.3 | c.3080A>G | p.(His1027Arg) |
| 17 | 59760676 | A | T | *BRIP1* | NM\_032043.2 | c.3731T>A | p.(Met1244Lys) |
| SAYO 019125 | 1 | 45797860 | C | T | *MUTYH* | NM\_001048171.1 | c.869G>A | p.(Ser290Asn) |
| SAYO 019133 | 5 | 236628 | C | T | *SDHA* | NM\_004168.3 | c.1346C>T | p.(Ala449Val) |
| 9 | 97864059 | A | G | *FANCC* | NM\_000136.2 | c.1607T>C | p.(Leu536Pro) |
| 12 | 58145346 | C | T | *CDK4* | NM\_000075.3 | c.155G>A | p.(Ser52Asn) |
| SAYO 019135 | 3 | 14200115 | C | T | *XPC* | NM\_001145769.1 | c.1157G>A | p.(Arg386Gln) |
| 8 | 1.46E+08 | C | T | *RECQL4* | NM\_004260.3 | c.2087G>A | p.(Arg696His) |
| SAYO 019136 | 1 | 45796909 | C | T | *MUTYH* | NM\_001048171.1 | c.1379G>A | p.(Arg460His) |
| 12 | 1.33E+08 | C | G | *POLE* | NM\_006231.3 | c.3140G>C | p.(Gly1047Ala) |
| SAYO 019137 | 11 | 1.08E+08 | A | G | *ATM* | NM\_000051.3 | c.185+3A>G | p.? |
| 19 | 50905076 | G | C | *POLD1* | NM\_001256849.1 | c.358G>C | p.(Gly120Arg) |
| SAYO 019141 | 12 | 1.33E+08 | C | T | *POLE* | NM\_006231.3 | c.3245G>A | p.(Arg1082His) |
| 16 | 68844167 | T | C | *CDH1* | NM\_004360.4 | c.755T>C | p.(Val252Ala) |
| SAYO 019142 | 19 | 11141459 | G | A | *SMARCA4* | NM\_001128844.1 | c.3436G>A | p.(Gly1146Ser) |
| SAYO 019143 | 22 | 29091782 | G | A | *CHEK2* | NM\_001005735.1 | c.1304C>T | p.(Ala435Val) |
| SAYO 019144 | 11 | 1.08E+08 | A | G | *ATM* | NM\_000051.3 | c.8734A>G | p.(Arg2912Gly) |
| 15 | 91295064 | A | G | *BLM* | NM\_000057.3 | c.847A>G | p.(Thr283Ala) |
| SAYO 019149 | 4 | 55161348 | T | A | *PDGFRA* | NM\_006206.5 | c.3179T>A | p.(Ile1060Asn) |
| SAYO 019152 | 10 | 43622132 | G | A | *RET* | NM\_020630.4 | c.3149G>A | p.(Arg1050Gln) |
| SAYO 019153 | 7 | 55227923 | T | A | *EGFR* | NM\_005228.4 | c.1390T>A | p.(Ser464Thr) |
| 8 | 30948047 | T | G | *WRN* | NM\_000553.4 | c.1719T>G | p.(Thr573=) |
| SAYO 019159 | 2 | 48028048 | C | T | *MSH6* | NM\_000179.2 | c.2926C>T | p.(Arg976Cys) |
| 12 | 1.33E+08 | G | A | *POLE* | NM\_006231.3 | c.3229C>T | p.(Arg1077Cys) |
| SAYO 19160 | 2 | 47630335 | C | T | *MSH2* | NM\_000251.2 | c.5C>T | p.(Ala2Val) |
| SAYO 019162 | 19 | 50910593 | G | A | *POLD1* | NM\_001256849.1 | c.1696G>A | p.(Glu566Lys) |
| SAYO 019163 | 19 | 1226569 | C | T | *STK11* | NM\_000455.4 | c.1225C>T | p.(Arg409Trp) |
| SAYO 019164 | 2 | 29917860 | A | G | *ALK* | NM\_004304.4 | c.808T>C | p.(Phe270Leu) |
| SAYO 019167 | 2 | 47630350 | A | G | *MSH2* | NM\_000251.2 | c.20A>G | p.(Glu7Gly) |
| 2 | 1.28E+08 | C | A | *ERCC3* | NM\_000122.1 | c.2207G>T | p.(Arg736Ile) |
| SAYO 019170 | 2 | 1.28E+08 | C | T | *ERCC3* | NM\_000122.1 | c.2228G>A | p.(Arg743His) |
| SAYO 020209 | 3 | 128200785 | C | T | GATA2 | [NM\_032638.5](http://www.ncbi.nlm.nih.gov/nuccore/NM_032638.5) | c.1020G>A | p.= |
| SAYO 020215 | 16 | 89849480 | C | T | FANCA | NM\_000135.2 | c.1501G>A | p.(Gly501Ser) |
| SAYO 020204 | 2 | 48023171 | C | T | MSH6 | NM\_000179.2 | c.596C>T | p.(Pro199Leu) |
| 8 | 145736926 | T | C | RECQL4 | NM\_004260.3 | c.3515A>G | p.(Tyr1172Cys) |
| SAYO 20218 | 2 | 47657058 | A | G | MSH2 | NM\_000251.2 | c.1254A>G | p.(Ile418Met) |
| 2 | 232995393 | A | C | DIS3L2 | [NM\_152383.5](http://www.ncbi.nlm.nih.gov/nuccore/NM_152383.5) | c.666A>C | p.(Arg222Ser) |
| 9 | 98270607 | G | C | PTCH1 | [NM\_000264.5](http://www.ncbi.nlm.nih.gov/nuccore/NM_000264.5) | c.37C>G | p.(Arg13Gly) |
| SAYO 020202 | 8 | 145738720 | C | T | RECQL4 | NM\_004260.3 | c.2344G>A | p.(Asp782Asn) |
| SAYO 019180 | 15 | 91304000 | G | A | BLM | NM\_000057.3 | c.1397G>A | p.(Gly466Glu) |
| 19 | 1207006 | A | G | STK11 | NM\_000455.4 | c.94A>G | p.(Thr32Ala) |
| 19 | 33792731 | GGCGGGT | G | CEBPA | NM\_004364.4 | c.584\_589del | p.(His195\_Pro196del) |
| SAYO 019197 | 11 | 108183193 | AAAAGT | A | ATM | NM\_000051.3 | c.5979\_5983del | p.(Ser1993Argfs\*23) |
| 16 | 2130208 | C | T | TSC2 | [NM\_000548.5](http://www.ncbi.nlm.nih.gov/nuccore/NM_000548.5) | c.3440C>T | p.(Ser1147Phe) |
| 16 | 23646911 | G | A | PALB2 | [NM\_024675.4](http://www.ncbi.nlm.nih.gov/nuccore/NM_024675.4) | c.956C>T | p.(Ser319phe) |
| 16 | 89865593 | G | C | FANCA | NM\_000135.2 | c.874C>G | p.(His292Asp) |
| SAYO 019196 | 3 | 14220032 | C | G | XPC | NM\_001145769.1 | c.37G>C | p.(Gly13Arg) |
| SAYO 019193 | 1 | 45798269 | T | C | MUTYH | NM\_001048171.1 | c.625A>G | p.(Ile209Val) |
| SAYO 019187 | 8 | 145738508 | C | T | RECQL4 | NM\_004260.3 | c.2477G>A | p.(Arg826Gln) |

Chr: Chromosome; Ref: Reference allele; Alt: Alternative allele; and AA: Amino acid