

HCP panel Gene List	Location	Gene Information
<u>APC</u>	5q22.2	adenomatous polyposis coli (APC) gene is a tumour suppressor gene which protein helps control cell growth and division. Pathogenic variants in the APC gene cause familial adenomatous polyposis 1. The risk of pancreatic cancer is also increased compared to the general population.
<u>ATM</u>	11q22.3	ataxia telangiectasia mutated (ATM) gene protein is involved in cell division and DNA repair. Changes in the DNA code which shortens the coding sequence of the <i>ATM</i> gene can lead to an increased risk of cancer including breast, ovarian and prostate.
<u>BAP1</u>	3p21.1	BRCA1 associated protein 1 (BAP1) gene is a tumour suppressor gene which produces a protein involved in cell growth, division and cell death. Pathogenic variants in the <i>BAP1</i> gene cause BAP1 tumour predisposition syndrome. An increased risk of melanoma and renal cancer have also been associated with risk increasing variants in <i>BAP1</i> .
<u>BARD1</u>	2q35	BRCA1-associated RING domain-1 (BARD1) gene is thought to play a role in <i>BRCA1</i> mediated tumour suppression. The protein encoded by <i>BARD1</i> interacts with <i>BRCA1</i> (breast cancer 1) protein. Pathogenic variants in <i>BARD1</i> can disrupt this interaction with <i>BRCA1</i> , which can lead to an increased risk of breast and ovarian cancer.
<u>BMPR1A</u>	10q23.2	bone morphogenetic protein receptor type 1A (BMPR1A) . Pathogenic variations in the <i>BMPR1A</i> gene can cause hereditary mixed polyposis syndrome-2 associated with and increased risk of colorectal cancer.
<u>BRCA1</u>	17q21.31	breast cancer 1 (BRAC1) gene produces a tumour suppressor protein which helps control cell growth and division along with DNA damage repair. Pathogenic variation in this gene have been associated with an increased risk of breast cancer, the risk for ovarian, prostate and pancreatic cancer is also increased. Pathogenic variants in <i>BRCA1</i> and <i>BRCA2</i> account for 5-10% of all breast cancers detected.
<u>BRCA2</u>	13q13.1	breast cancer 2 (BRCA2) gene produces a tumour suppressor protein which helps control cell growth and division along with DNA damage repair. Pathogenic variation in this gene have been associated with an increased risk of breast and ovarian cancer, there is also a smaller increased risk of melanoma, prostate and pancreatic cancer. Pathogenic variants in <i>BRCA1</i> and <i>BRCA2</i> account for 5-10% of all breast cancers detected.
<u>BRIP1</u>	17q23.2	BRCA1-interacting protein 1 (BRIP1) interacts with <i>BRCA1</i> protein to repair damaged DNA. Pathogenic variants in this gene have been linked to ovarian cancer and a smaller increased risk of breast cancer.
<u>CDH1</u>	16q22.1	cadherin 1 (CDH1) gene produces the protein E-cadherin which acts as a tumour suppressor protein preventing uncontrolled cell growth and division. Cancers associated with pathogenic variants in this gene include gastric cancer and lobular breast cancers.
<u>CDK4</u>	12q14.1	cyclin dependent kinase-4 (CDK4) gene mutations are associated with an increased risk of cutaneous malignant melanoma-3.
<u>CDKN2A</u>	9p21.3	cyclin dependent kinase inhibitor 2A (CDKN2A) produces a protein which function as tumour suppressors. Cancers associated with pathogenic variants in the <i>CDKN2A</i> gene include melanoma and pancreatic cancer.

<u>CHEK2</u>	22q12.1	checkpoint kinase-2 (CHEK2) gene produces a protein involved in cell cycle arrest and DNA repair when DNA is damaged. Changes in the DNA code which shortens the coding sequence of the CHEK2 gene can lead to an increased risk of cancer including breast, ovarian and prostate.
<u>EPCAM</u>	2p21	epithelial cell adhesion molecule (EPCAM) gene produces a protein involved in the regulation of gene activity of many genes involved in cellular processes including cell growth and division. Pathogenic variants in <i>EPCAM</i> are associated with Lynch syndrome. Lynch syndrome is a condition which increases the risk of developing certain cancers including breast, colon, pancreatic, prostate, ovarian and uterine.
<u>HOXB13</u>	17q21.32	homeobox B13 (HOXB13) gene produces a tumour suppressor protein which helps control cell growth and division. Pathogenic variants in the <i>HOXB13</i> gene have been associated with an increased risk to prostate cancer.
<u>MLH1</u>	3p22.2	mutL homolog 1 (MLH1) gene protein is essential to repairing errors in DNA replication. MLH1 gene is a mismatch repair gene. Pathogenic variants in MLH1 are associated with Lynch syndrome. Lynch syndrome is a condition which increases the risk of developing certain cancers including breast, colon, pancreatic, prostate and ovarian.
<u>MSH2</u>	2p21-p16.3	mutS homolog 2 (MSH2) gene protein is essential to repairing errors in DNA replication. <i>MSH2</i> gene is a mismatch repair gene. Pathogenic variants in MSH2 are associated with Lynch syndrome. Lynch syndrome is a condition which increases the risk of developing certain cancers including breast, colon, pancreatic, prostate, ovarian and uterine.
<u>MSH6</u>	2p16.3	mutS homolog 6 (MSH6) gene protein is essential to repairing errors in DNA replication. <i>MSH6</i> gene is a mismatch repair gene. Pathogenic variants in <i>MSH6</i> are associated with Lynch syndrome. Lynch syndrome is a condition which increases the risk of developing certain cancers including breast, colon, pancreatic, prostate, ovarian and uterine.
<u>MUTYH</u>	1p34.1	mutY DNA glycosylase (MUTYH) gene produces a protein involved in DNA repair. Some variations in the <i>MUTYH</i> gene cause familial adenomatous polyposis which increases the risk of colon cancer. <i>MUTYH</i> is an autosomal recessive gene. If both copies of the <i>MUTYH</i> gene are found to have a risk increasing variant, then the risk of colorectal cancer is increased.
<u>NF1</u>	17q11.2	neurofibromin 1 (NF1) gene produces a tumour suppressor protein. Neurofibromatosis Type 1 is caused by pathogenic variation in the <i>NF1</i> gene, and an increased risk of some cancers.
<u>NTHL1</u>	16p13.3	nth like DNA glycosylase 1 (NTHL1) is a base excision repair gene. Pathogenic variants in the <i>NTHL1</i> gene cause familial adenomatous polyposis-3 which increases the risk of colon cancer.
<u>PALB2</u>	16p12.2	partner and localizer of BRCA2 (PALB2) gene protein is involved in recombination repair and cell cycle checkpoints. Pathogenic variants in <i>PALB2</i> can be associated with an increased risk of breast, ovarian and pancreatic cancer.
<u>PMS2</u>	7p22.1	PMS1 homolog 2 (PMS2) gene protein is essential to repairing errors in DNA replication. <i>PMS2</i> gene is a mismatch repair gene. Lynch syndrome is associated with mutations in PMS2 Pathogenic variants in <i>PMS2</i> are associated with Lynch syndrome. Lynch syndrome is a condition which increases the risk of developing certain cancers including colon cancer.
<u>POLD1</u>	19q13.33	DNA polymerase-delta (POLD1) gene protein is involved in DNA replication and repair. Some variants in <i>POLD1</i> increase the risk of colorectal cancer.
<u>POLE</u>	12q24.33	DNA polymerase-epsilon (POLE) gene protein is involved in DNA replication and repair. Some variants in <i>POLE</i> increase the risk of colorectal cancer.

<u>PTEN</u>	10q23.31	phosphatase and tensin homolog (<i>PTEN</i>) gene encodes a tumour suppressor gene protein. Many cancers have been associated with pathogenic variants in the <i>PTEN</i> gene including breast, ovarian and prostate cancer. Cowden syndrome characterised by an increased risk of developing certain cancers in caused by mutations in <i>PTEN</i> .
<u>RAD51C</u>	17q22	RAD51 paralog C (<i>RAD51C</i>) gene produces a protein involved in homologous recombination and DNA repair. Ovarian cancer has been associated with mutations in the <i>RAD51C</i> gene.
<u>RAD51D</u>	17q12	RAD51 paralog D (<i>RAD51D</i>) gene produces a protein involved in homologous recombination and DNA repair. Ovarian cancer has been associated with mutations in the <i>RAD51D</i> gene.
<u>SMAD4</u>	18q21.2	SMAD family member 4 (<i>SMAD4</i>) gene produces a protein involved in the signalling pathway. The <i>SMAD4</i> protein is a transcription factor and tumour suppressor. Pathogenic variations in the <i>SMAD4</i> gene have been associated with an increased risk of colorectal cancer.
<u>STK11</u>	19p13.3	serine/threonine kinase 11 (<i>STK11</i>) gene produces a tumour suppressor enzyme which controls cell growth and division. Pathogenic variation in the <i>STK11</i> gene cause Peutz-Jeghers syndrome, a condition characterised by hamartomatous polyps in the gastrointestinal tract and an increased risk of developing several types of cancer.
<u>TP53</u>	17p13.1	tumour protein 53 (<i>TP53</i>) gene encodes the protein tumour protein p53, a tumour suppressor protein involved in regulating DNA repair and cell division. Li Fraumeni Syndrome is associated with pathogenic variation in <i>TP53</i> , which increases the risk of developing many types of soft tissue cancers at a younger age.
<u>VHL</u>	3p25.3	von Hippel-Lindau tumour suppressor (<i>VHL</i>) gene plays a role tumour suppression and formation of the extracellular matrix. Von Hippel-Lindau syndrome is caused by pathogenic variation in the <i>VHL</i> gene and characterised by the formation of tumours and cysts.
