HCP panel Gene List	Location	Gene Information
APC	**5q22.2**	**Adenomatous polyposis coli *(APC)*** The *APC* tumour suppressor gene encodes the APC protein which 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.
ATM	**11q22.3**	**Ataxia telangiectasia mutated *(ATM)*** The *ATM* gene encodes a protein involved in cell division and DNA repair. Changes in the DNA code which shortens the coding sequence of the *ATM* gene can lead to an increased risk of cancer including breast, ovarian and prostate.
BAP1	**3p21.1**	**BRCA1 associated protein 1 *(BAP1)*** The *BAP1* gene is a tumour suppressor gene which produces a protein involved in cell growth, division and cell death. Pathogenic variants in the *BAP1* gene cause BAP1 tumour predisposition syndrome. An increased risk of melanoma and renal cancer have also been associated with pathogenic variants in *BAP1*.
BARD1	**2q35**	**BRCA1-associated RING domain-1 *(BARD1)*** The *BARD1* is thought to play a role in *BRCA1* mediated tumour suppression. The protein encoded by *BARD1* interacts with BRCA1 (breast cancer 1) protein. Pathogenic variants in *BARD1* can disrupt this interaction with BRCA1, which can lead to an increased risk of breast and ovarian cancer.
BMPR1A	**10q23.2**	**Bone morphogenetic protein receptor type 1A *(BMPR1A)*** Pathogenic variations in the *BMPR1A* gene can cause hereditary mixed polyposis syndrome-2 associated with and increased risk of colorectal cancer.
BRCA1	**17q21.31**	**Breast cancer 1 *(BRAC1)*** The *BRAC1* gene produces a tumour suppressor protein which helps control cell growth and division along with DNA damage repair. Pathogenic variation in this gene has been associated with an increased risk of breast cancer, the risk for ovarian, prostate and pancreatic cancer is also increased. Pathogenic variants in *BRCA1* and *BRCA2* account for 5-10% of all breast cancers detected.
BRCA2	**13q13.1**	**Breast cancer 2 *(BRCA2)*** The *BRACA2* gene produces a tumour suppressor protein which helps control cell growth and division along with DNA damage repair. Pathogenic variation in this gene has 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 *BRCA1* and *BRCA2* account for 5-10% of all breast cancers detected.
BRIP1	**17q23.2**	**BRCA1-interacting protein 1 *(BRIP1)*** *BRIP1* interacts with BRCA1 protein to repair damaged DNA. Pathogenic variants in this gene have been linked to ovarian cancer and a smaller increased risk of breast cancer.
CDH1	**16q22.1**	**Cadherin 1 *(CDH1)*** *CDH1* 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.

CDK4	**12q14.1**	**Cyclin dependent kinase-4 *(CDK4)*** Pathogenic variants in the *CDK4* gene are associated with an increased risk of cutaneous malignant melanoma-3.
CDKN2A	**9p21.3**	**Cyclin dependent kinase inhibitor 2A *(CDKN2A)*** *CDKN2A* is a tumour suppressor gene. Cancers associated with pathogenic variants in the *CDKN2A* gene include melanoma and pancreatic cancer.
CHEK2	**22q12.1**	**Checkpoint kinase-2 *(CHEK2)*** *CHEK2* 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.
EPCAM	**2p21**	**Epithelial cell adhesion molecule (EPCAM)** *EPCAM* 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 *EPCAM* 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.
HOXB13	**17q21.32**	**Homeobox B13 *(HOXB13)*** *HOXB13* is a tumour suppressor gene which helps control cell growth and division. Pathogenic variants in the *HOXB13* gene have been associated with an increased risk to prostate cancer.
MLH1	**3p22.2**	**MutL homolog 1 *(MLH1)*** *MLH1* gene protein in essential to repairing errors in DNA replication. *MLH1* 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.
MSH2	**2p21-p16.3**	**MutS homolog 2 *(MSH2)*** *MSH2* gene protein in essential to repairing errors in DNA replication. *MSH2* 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.
MSH6	**2p16.3**	**MutS homolog 6 (*MSH6*)** *MSH6* gene protein in essential to repairing errors in DNA replication. *MSH6* is a mismatch repair gene. Pathogenic variants in *MSH6* 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.
MUTYH	**1p34.1**	**MutY DNA glycosylase *(MUTYH)*** *MUTYH* produces a protein involved in DNA repair. Some variations in *MUTYH* cause familial adenomatous polyposis which increases the risk of colon cancer. *MUTYH* is an autosomal recessive gene. If both copies of the *MUTYH* gene are found to have a risk increasing variant, then the risk of colorectal cancer is increased.
NF1	**17q11.2**	**Neurofibromin 1 *(NF1)*** *NF1* produces a tumour suppressor protein. Neurofibromatosis Type 1 is caused by pathogenic variation in the *NF1* gene, and an increased risk of some cancers.
NTHL1	**16p13.3**	**Nth like DNA glycosylase 1 *(NTHL1)*** Is a base excision repair gene. Pathogenic variants in the *NTHL1* gene cause familial adenomatous polyposis-3 which increases the risk of colon cancer.

PALB2	**16p12.2**	**Partner and localizer of BRCA2 *(PALB2)*** *PALB2* gene protein is involved in recombination repair and cell cycle checkpoints. Pathogenic variants in *PALB2* can be associated with an increased risk of breast, ovarian and pancreatic cancer.
PMS2	**7p22.1**	**PMS1 homolog 2 *(PMS2)*** *PMS2* gene protein in essential to repairing errors in DNA replication. *PMS2* is a mismatch repair gene. Lynch syndrome is associated with mutations in *PMS2*. Pathogenic variants in *PMS2* are associated with Lynch syndrome. Lynch syndrome is a condition which increases the risk of developing certain cancers including colon cancer.
POLD1	**19q13.33**	**DNA polymerase-delta *(POLD1)*** *POLD1* gene protein is involved in DNA replication and repair. Some variants in *POLD1* increase the risk of colorectal cancer.
POLE	**12q24.33**	**DNA polymerase-epsilon (POLE)** *POLE* gene protein is involved in DNA replication and repair. Some variants in *POLE* increase the risk of colorectal cancer.
PTEN	**10q23.31**	**Phosphatase and tensin homolog *(PTEN)*** *PTEN* is a tumour suppressor gene. Many cancers have been associated with pathogenic variants in the *PTEN* gene including breast, ovarian and prostate cancer. Cowden syndrome characterised by an increased risk of developing certain cancers in caused by pathogenic variants in *PTEN*.
RAD51C	**17q22**	**RAD51 paralog C *(RAD51C)*** *RAD51C* produces a protein involved in homologous recombination and DNA repair. Breast and ovarian cancer has been associated with mutations in the *RAD51C* gene.
RAD51D	**17q12**	**RAD51 paralog D *(RAD51D)*** *RAD51D* produces a protein involved in homologous recombination and DNA repair. Breast and ovarian cancer has been associated with mutations in the *RAD51D* gene.
SMAD4	**18q21.2**	**SMAD family member 4 *(SMAD4)*** *SMAD4* produces a protein involved in the signalling pathway. The SMAD4 protein is a transcription factor and tumour suppressor. Pathogenic variations in the *SMAD4* gene have been associated with an increased risk of colorectal cancer.
STK11	**19p13.3**	**Serine/threonine kinase 11 (STK11)** *STK11* produces a tumour suppressor enzyme which controls cell growth and division. Pathogenic variation in the *STK11* 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.
TP53	**17p13.1**	**Tumour protein 53 *(TP53)*** *TP53* encodes the tumour suppressor protein p53, involved in regulating DNA repair and cell division. Li Fraumeni Syndrome is associated with pathogenic variation in *TP53*, which increases the risk of developing many types of soft tissue cancers at a younger age.
VHL	**3p25.3**	**Von Hippel-Lindau tumour suppressor *(VHL)*** *VHL* plays a role tumour suppression and formation of the extracellular matrix. Von Hippel-Lindau syndrome is caused by pathogenic variation in the *VHL* gene and characterised by the formation of tumours and cysts.