List of diseases covered by Cancer Predisposition NGS panel

Gene	Condition
AIP	Pituitary adenoma 1, multiple types
ALK	Neuroblastoma, susceptibility to, 3
APC	Adenomatous polyposis coli; Colorectal cancer, somatic; Desmoid disease, hereditary; Gardner syndrome; Gastric cancer, somatic; Hepatoblastoma, somatic
ATM	Ataxia-telangiectasia; Breast cancer, susceptibility to
BAP1	Tumor predisposition syndrome
BARD1	Breast cancer, susceptibility to
BLM	Bloom syndrome
BMPR1A	Juvenile polyposis syndrome, infantile form; Polyposis syndrome, hereditary mixed, 2
BRCA1	Breast-ovarian cancer, familial, 1; Pancreatic cancer, susceptibility to, 4; Fanconi anemia, complementation group S
BRCA2	Fanconi anemia, complementation group D1; Wilms tumor; Breast cancer, male, susceptibility to; Breast-ovarian cancer, familial, 2; Glioblastoma 3; Medulloblastoma; Pancreatic cancer 2; Prostate cancer
BRIP1	Breast cancer, early-onset; Fanconi anemia, complementation group J
BUB1B	Colorectal cancer, somatic; Mosaic variegated aneuploidy syndrome 1; Premature chromatid separation trait; Colorectal cancer, somatic
CDC73	Hyperparathyroidism, familial primary; Hyperparathyroidism-jaw tumor syndrome; Parathyroid carcinoma
CDH1	Breast cancer, lobular; Prostate cancer, susceptibility to; Endometrial carcinoma, somatic; Ovarian carcinoma, somatic; Blepharocheilodontic syndrome 1; Gastric cancer, familial diffuse, with or without cleft lip and/or palate
CDK4	Melanoma, cutaneous malignant, 3
CDKN1C	

Beckwith-Wiedemann syndrome; IMAGE syndrome
CDKN2A
Melanoma and neural system tumor syndrome;
Pancreatic cancer/melanoma syndrome;
Melanoma, cutaneous malignant, 2
CEBPA
Leukemia, acute myeloid
CEP57
Mosaic variegated aneuploidy syndrome 2
CHEK2
Breast cancer, susceptibility to; Prostate cancer, familial, susceptibility to;
Li-Fraumeni syndrome; Osteosarcoma, somatic
CYLD
Trichoepithelioma, multiple familial, 1; Brooke-Spiegler syndrome;
Cylindromatosis, familial
DDB2
Xeroderma pigmentosum, group E, DDB-negative subtype
DICER1
Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors;
Pleuropulmonary blastoma; Rhabdomyosarcoma, embryonal, 2
DIS3L2
Perlman syndrome
EGFR
Adenocarcinoma of lung, response to tyrosine kinase inhibitor in
EPCAM
Colorectal cancer, hereditary nonpolyposis, type 8;
Diarrhea 5, with tufting enteropathy, congenital
ERCC2
Xeroderma pigmentosum, group D;
Cerebrooculofacioskeletal syndrome 2;
Trichothiodystrophy 1, photosensitive
ERCC3
Xeroderma pigmentosum, group B;
Trichothiodystrophy 2, photosensitive
ERCC4
Xeroderma pigmentosum, group F;
Fanconi anemia, complementation group Q;
XFE progeroid syndrome
ERCC5
Xeroderma pigmentosum, group G;
Cerebrooculofacioskeletal syndrome 3
EXT1
Chondrosarcoma; Exostoses, multiple, type 1
EXT2
Exostoses, multiple, type 2;
Seizures, scoliosis, and macrocephaly syndrome
EZH2
Weaver syndrome
FANCA
Fanconi anemia, complementation group A
FANCB
Fanconi anemia, complementation group B

FANCC
Fanconi anemia, complementation group C
FANCD2
Fanconi anemia, complementation group D2
FANCE
Fanconi anemia, complementation group E
FANCF
Fanconi anemia, complementation group F
FANCG
Fanconi anemia, complementation group G
FANCI
Fanconi anemia, complementation group I
FANCL
Fanconi anemia, complementation group L
FH
Leiomyomatosis and renal cell cancer; Fumarase deficiency
FLCN
Birt-Hogg-Dube syndrome; Pneumothorax, primary spontaneous;
Colorectal cancer, somatic; Renal carcinoma, chromophobe, somatic
GATA2
Emberger syndrome; Immunodeficiency 21;
Leukemia, acute myeloid, susceptibility to;
Myelodysplastic syndrome, susceptibility to
GPC3
Simpson-Golabi-Behmel syndrome, type 1; Wilms tumor, somatic
HNF1A
Renal cell carcinoma; Hepatic adenoma, somatic;
Diabetes mellitus, insulin-dependent, 20; MODY, type III;
Diabetes mellitus, insulin-dependent;
Diabetes mellitus, noninsulin-dependent, 2
HRAS
Costello syndrome; Schimmelpenning-Feuerstein-Mims syndrome,
somatic mosaic; Bladder cancer, somatic;
Nevus sebaceous or woolly hair nevus, somatic;
Spitz nevus or nevus spilus, somatic; Thyroid carcinoma, follicular, somatic
KIT
Gastrointestinal stromal tumor, familial; Germ cell tumors, somatic;
Leukemia, acute myeloid; Mastocytosis, cutaneous; Piebaldism
MAX
Pheochromocytoma, susceptibility to
MEN1
Multiple endocrine neoplasia 1
MET
Hepatocellular carcinoma, childhood type, somatic;
Renal cell carcinoma, papillary, 1, familial and somatic;
Deafness, autosomal recessive 97; Osteofibrous dysplasia, susceptibility to
MITF
Melanoma, cutaneous malignant, susceptibility to, 8;
COMMAD syndrome; Tietz albinism-deafness syndrome;
Waardenburg syndrome, type 2A;
Waardenburg syndrome/ocular albinism, digenic
MLH1

Mismatch repair cancer syndrome;
Colorectal cancer, hereditary nonpolyposis, type 2; Muir-Torre syndrome
MRE11A
Ataxia-telangiectasia-like disorder 1
MSH2
Mismatch repair cancer syndrome;
Colorectal cancer, hereditary nonpolyposis, type 1; Muir-Torre syndrome
MSH6
Mismatch repair cancer syndrome;
Colorectal cancer, hereditary nonpolyposis, type 5;
Endometrial cancer, familial
MUTYH
Adenomas, multiple colorectal;
Colorectal adenomatous polyposis, autosomal recessive,
with pilomatricomas; Gastric cancer, somatic
NBN
Nijmegen breakage syndrome; Leukemia, acute lymphoblastic;
Aplastic anemia
NF1
Neurofibromatosis, type 1
NF2
Meningioma, NF2-related, somatic; Neurofibromatosis, type 2;
Schwannomatosis, somatic
NSD1
Leukemia, acute myeloid; Sotos syndrome 1
NTHL1
Familial adenomatous polyposis 3
PALB2
Breast cancer, susceptibility to;
Pancreatic cancer, susceptibility to, 3;
Fanconi anemia, complementation group N
PHOX2B
Neuroblastoma with Hirschsprung disease;
Central hypoventilation syndrome, congenital,
with or without Hirschsprung disease
POLD1
Colorectal cancer, susceptibility to, 10;
Mandibular hypoplasia, deafness, progeroid features,
and lipodystrophy syndrome
PRF1
Aplastic anemia; Hemophagocytic lymphohistiocytosis, familial, 2;
Lymphoma, non-Hodgkin
PRKAR1A
Acrodysostosis 1, with or without hormone resistance;
Carney complex, type 1; Myxoma, intracardiac;
Pigmented nodular adrenocortical disease, primary, 1
PRSS1
Pancreatitis, hereditary; Trypsinogen deficiency
PTCH1
Basal cell nevus syndrome; Holoprosencephaly 7;
Basal cell carcinoma, somatic
PTEN

Glioma susceptibility 2, Meningioma, Macrocephaly/autism syndrome;
Endometrial carcinoma, somatic; Malignant melanoma, somatic;
Squamous cell carcinoma, head and neck, somatic; Prostate cancer, somatic;
VATER association with macrocephaly and ventriculomegaly;
Bannayan-Riley-Ruvalcaba syndrome; Cowden syndrome 1
RAD50
Nijmegen breakage syndrome-like disorder
RAD51C
Breast-ovarian cancer, familial, susceptibility to, 3;
Fanconi anemia, complementation group O
RAD51D
Breast-ovarian cancer, familial, susceptibility to, 4
RB1
Bladder cancer, somatic; Osteosarcoma, somatic; Retinoblastoma;
Small cell cancer of the lung, somatic
RECQL4
Baller-Gerold syndrome; RAPADILINO syndrome;
Rothmund-Thomson syndrome
RET
Medullary thyroid carcinoma;
Central hypoventilation syndrome, congenital;
Multiple endocrine neoplasia IIA; Multiple endocrine neoplasia IIB;
Pheochromocytoma; Hirschsprung disease, susceptibility to, 1
RHBDF2
Tylosis with esophageal cancer
RUNX1
Leukemia, acute myeloid; Platelet disorder, familial,
with associated myeloid malignancy
SBDS
Shwachman-Diamond syndrome; Aplastic anemia, susceptibility to
SDHA
Cardiomyopathy, dilated, 1GG; Leigh syndrome;
Mitochondrial respiratory chain complex II deficiency; Paragangliomas 5
SDHAF2
Paragangliomas 2
SDHB
Cowden syndrome 2; Gastrointestinal stromal tumor;
Paraganglioma and gastric stromal sarcoma; Paragangliomas 4;
Pheochromocytoma
SDHC
Gastrointestinal stromal tumor;
Paraganglioma and gastric stromal sarcoma; Paragangliomas 3
SDHD
Carcinoid tumors, intestinal; Cowden syndrome 3;
Mitochondrial complex II deficiency;
Paraganglioma and gastric stromal sarcoma; Pheochromocytoma
SLX4
Fanconi anemia, complementation group P
SMAD4
Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;
Myhre syndrome; Pancreatic cancer, somatic; Polyposis, juvenile intestinal
SMARCB1

Coffin-Siris syndrome 3;
Rhabdoid tumor predisposition syndrome 1;
Schwannomatosis-1, susceptibility to
STK11
Pancreatic cancer; Peutz-Jeghers syndrome;
Testicular tumor, somatic
SUFU
Basal cell nevus syndrome; Joubert syndrome 32;
Medulloblastoma, desmoplastic; Meningioma, familial, susceptibility to
TMEM127
Pheochromocytoma, susceptibility to
TP53
Breast cancer; Adrenal cortical carcinoma;
Choroid plexus papilloma;
Colorectal cancer; Li-Fraumeni syndrome;
Nasopharyngeal carcinoma; Osteosarcoma; Pancreatic cancer;
Basal cell carcinoma 7;
Glioma susceptibility 1
TSC1
Focal cortical dysplasia, type II, somatic;
Lymphangioleiomyomatosis; Tuberous sclerosis-1
TSC2
Focal cortical dysplasia, type II, somatic;
Lymphangioleiomyomatosis, somatic; Tuberous sclerosis-2
VHL
Erythrocytosis, familial, 2; Pheochromocytoma;
Renal cell carcinoma, somatic; von Hippel-Lindau syndrome
WRN
Werner syndrome
WT1
Wilms tumor, type 1; Denys-Drash syndrome;
Frasier syndrome; Meacham syndrome; Mesothelioma, somatic;
Nephrotic syndrome, type 4
XPA
Xeroderma pigmentosum, group A
XPC
Xeroderma pigmentosum, group C
XRCC2
Fanconi anemia, complementation group U