UPMC
LIFECHANGINGMEDICINE

PITTSBURGH CYTOGENETICS LABORATORY

CANCER SYNDROMES COVERED BY KANCERRAY

Gene	Chromosome	Cancer Syndrome
ALK	2p23	Familial neuroblastoma
APC	5q21	Adenomatous polyposis coli; Turcot syndrome
ATM	11q22.3	Ataxia-telangiectasia
BLM	15q26.1	Bloom Syndrome
BMPR1A	10q22.3	Juvenile polyposis
BRCA1	17q21	Hereditary breast/ovarian cancer
BRCA2	13q12	Hereditary breast/ovarian cancer
BRIP1	17q22	Fanconi anaemia J, breast cancer susceptiblity
BUB1B	15q15	Mosaic variegated aneuploidy
CDC73	1q21-q31	Hyperparathyroidism-jaw tumor syndrome
CDH1	16q22.1	Familial gastric carcinoma
CDK4	12q14	Familial malignant melanoma
CDKN2A	9p21	Familial malignant melanoma
CHEK2	22q12.1	familial breast cancer
CYLD	16q12-q13	Familial cylindromatosis
DDB2	11p12	Xeroderma pigmentosum (E)
DICER1	14q32.13	Familial Pleuropulmonary Blastoma
EGFR	7p12.3-p12.1	Familial lung cancer
ERCC2	19q13.2-q13.3	Xeroderma pigmentosum (D)
ERCC3	2q21	Xeroderma pigmentosum (B)
ERCC4	16p13.3-p13.13	Xeroderma pigmentosum (F)
ERCC5	13q33	Xeroderma pigmentosum (G)
EXT1	8q24.11-q24.13	Multiple Exostoses Type 1
EXT2	11p12-p11	Multiple Exostoses Type 2
FANCA	16q24.3	Fanconi anaemia A
FANCC	9q22.3	Fanconi anaemia C
FANCD2	3p26	Fanconi anaemia D2
FANCE	6p21-p22	Fanconi anaemia E
FANCF	11p15	Fanconi anaemia F
FANCG	9p13	Fanconi anaemia G
FH	1q42.1	hereditary leiomyomatosis and renal cell cancer
FLCN	17p11.2	Birt-Hogg-Dube syndrome
GPC3	Xq26.1	Simpson-Golabi-Behmel syndrome
HNF1A	12q24.2	Familial Hepatic Adenoma
HRAS	11p15.5	Costello syndrome
KIT	4q12	Familial gastrointestinal stromal tumour
MEN1	11q13	Multiple Endocrine Neoplasia Type 1
MET	7q31	Familial Papillary Renal Cancer
MLH1	3p21.3	Hereditary non-polyposis colorectal cancer, Turcot syndrome
MPL	p34	Familial essential thrombocythemia
MSH2	2p22-p21	Hereditary non-polyposis colorectal cancer
MSH6	2p16	Hereditary non-polyposis colorectal cancer
MUTYH	1p34.3-1p32.1	Adenomatous polyposis coli

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Gene	Chromosome	Cancer Syndrome
NBN	8q21	Nijmegen breakage syndrome
NF1	17q12	Neurofibromatosis type 1
NF2	22q12.2	Neurofibromatosis type 2
PALB2	16p12.1	Fanconi anaemia N, breast cancer susceptibility
PHOX2B	4p12	familial neuroblastoma
PMS1	2q31-q33	Hereditary non-polyposis colorectal cancer
PMS2	7p22	Hereditary non-polyposis colorectal cancer, Turcot syndrome
PRKAR1A	17q23-q24	Carney complex
PTCH1	9q22.3	Nevoid Basal Cell Carcinoma Syndrome
PTEN	10q23.3	Cowden Syndrome, Bannayan-Riley-Ruvalcaba syndrome
RB1	13q14	Familial retinoblastoma
RECQL4	8q24.3	Rothmund-Thompson Syndrome
RET	10q11.2	Multiple endocrine neoplasia 2A/2B
SBDS	7q11	Schwachman-Diamond syndrome
SDHAF2	11q12.2	Familial paraganglioma
SDHB	1p36.1-p35	Familial paraganglioma
SDHC	1q21	Familial paraganglioma
SDHD	11q23	Familial paraganglioma
SMAD4	18q21.1	Juvenile polyposis
SMARCB1	22q11	Rhabdoid predisposition syndrome
STK11	19p13.3	Peutz-Jeghers syndrome
SUFU	10q24.32	Medulloblastoma predisposition
TP53	17p13	Li-Fraumeni syndrome
TSC1	9q34	Tuberous sclerosis 1
TSC2	16p13.3	Tuberous sclerosis 2
VHL	3p25	von Hippel-Lindau syndrome
WAS	Xp11.23-p11.22	Wiskott-Aldrich syndrome
WRN	8p12-p11.2	Werner Syndrome
WT1	11p13	Denys-Drash syndrome, Frasier syndrome, Familial Wilms tumor
XPA	9q22.3	Xeroderma pigmentosum (A)
XPC	3p25	Xeroderma pigmentosum (C)

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