Genes classified under Hereditary Cancer [Source : Huang KL, Mashl RJ, Wu Y, et al. Pathogenic Germline Variants in 10,389 Adult Cancers. Cell. 2018;173(2):355-370.e14. doi:10.1016/j.cell.2018.03.039]

Gene Symbol	Gene Name	Gene Alias(es)	Chromosome location	Cancer syndrome(s)	Major associated tumor types	Mode of inheritance	Gene Start (bp)	Gene End (bp)	Reference (PubMed ID)	OMIM ID	Source
ATR	ATR serine/threonine kinase	ATR,Q13535,FRP1,SCKL1,SCKL,MEC1,ENSG00000175054,545	3q23	familial cutaneous telangiectasia and cancer sydrome, Seckel syndrome	endometrial, gastric	autosomal dominant	142449429	142578704	29025585	601215	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
FANCD2	Fanconi anemia, complementation group D2	2177,FANCD,DKFZp762A223,FAD2,FA-D2,FA4,FACD,FAD,FLJ23826,ENSG00000144554,FANCD2	3p26	Fanconi anaemia D2	AML, leukaemia	autosomal recessive	10028658	10098950	28678401	613984	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
FANCE	Fanconi anemia, complementation group E	FANCE,FACE,FAE,ENSG00000112039,2178	6p21-p22	Fanconi anaemia E	AML, leukaemia	autosomal recessive	35452546	35466345	28678401	613976	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
FANCF	Fanconi anemia, complementation group F	FANCF,FAF,MGC126856,Q9NPI8,ENSG00000183161,2188	11p15	Fanconi anaemia F	AML, leukaemia	autosomal recessive	22624686	22625810	28440438; 28687356	613897	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
HNF1A	HNF1 homeobox A	6927,HNF1,LFB1,P20823,HNF1A,MODY3,TCF1,ENSG00000135100,TCF1	12q24.2	familial hepatic adenoma	hepatic adenoma, hepatocellular carcinoma	autosomal recessive	120978769	121001192	23707370	142410	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
LMO1	LIM domain only 1 (rhombotin 1) (RBTN1)	LMO1,RBTN1,TTG1,RHOM1,P25800,ENSG00000166407,4004	11p15		neuroblastoma	autosomal dominant	8224616	8268634	21124317	186921	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
MPL	myeloproliferative leukaemia virus oncogene, thrombopoietin receptor	P40238,TPOR,RP1-92O14_1,C-MPL,MPLV,CD110,ENSG00000117400,4352, MPL	1p34	familial essential thrombocythemia	MPN	autosomal dominant	43337849	43352772	28979237	159530	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)

PMS1	PMS1 postmeiotic segregation increased 1 (S. cerevisiae)	5378,hPMS1,DKFZp781M0253,HNPCC3,PMSL1,ENSG00000064933,PMS1	2q31-q33		hereditary non-polyposis colorectal cancer	autosomal recessive	189791810	189877436	28687356	600258	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
PRF1	perforin 1 (pore forming protein)	P14222,PRF1,5551,ENSG00000180644,FLH2,HPLH2,MGC65093,P1,PFP	10q22		various leukaemia, lymphoma	autosomal recessive	70598053	70600902	29113160	170280	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
SETBP1	SET binding protein 1	Q9UEF3,SETBP1_ENST00000282030,ENSG00000152217,KIAA0437,SEB,26040,Q9Y6X0,A6H8W5,Q6P6C3	18q21.1	Schinzel-Giedion syndrome	neuroepithelial tumours	autosomal dominant	44701347	45063698	28383694	611060	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
TSHR	thyroid stimulating hormone receptor	7253,hTSHR-I,LGR3,MGC75129,ENSG00000165409,TSHR	14q31		thyroid adenoma	autosomal dominant	80955681	81144353	7920658	603372	Cancer Gene Census Germline download 1/5/2016 (http://cancer.sanger.ac.uk/census/)
DROSHA	RIBONUCLEASE III, NUCLEAR	RNASEN	5p13.3			autosomal dominant	314,00,493	315,32,174	personal communication; related to DICER1	608828	personal communication with Gang Wu, St. Jude; related to DICER1
ABCB11	ATP-binding cassette, sub-family B (MDR/TAP), member 11	ABC16, BSEP, PFIC-2, PFIC2, PGY4, SPGP	2q31.1	Progressive familial intrahepatic cholestasis	Hepatocellular carcinoma Cholangiocarcinoma	autosomal recessive	1697,79,448	1698,87,832	9806540	603201	Rahman 114 CPG
ALK	anaplastic lymphoma receptor tyrosine kinase	CD246, NBLST3	2p23		Neuroblastoma	autosomal dominant	294,15,640	301,44,432	18724359	105590	Rahman 114 CPG
APC	adenomatous polyposis coli	DP2.5	5q21	Familial adenomatous polyposis (FAP)	Colorectal cancer Hepatoblastoma Desmoid tumor	autosomal dominant	1120,43,195	1121,81,936	1651174 1651562 1651563 1678319	611731	Rahman 114 CPG

ATM	ataxia telangiectasia mutated	TEL1, TELO1	11q22.3	Ataxia-Telangiectasia (biallelic mutations)	Biallelic mutations: Lymphoid hematological malignancy (leukemia, lymphoma) Monoallelic mutations: Breast cancer	autosomal recessive autosomal dominant	1080,93,211	1082,39,829	7792600	607585	Rahman 114 CPG
AXIN2	axin 2	AXIL, ODCRCS	17q24.1	oligodentia-colorectal cancer syndrome	Colorectal cancer	autosomal dominant	6335,53,924	635,56,675	15042511	604025	Rahman 114 CPG
BAP1	BRCA1 associated protein-1 (ubiquitin carboxy-terminal hydrolase)	UCHL2	3p21.31-p21.2		Melanoma (cutaneous, uveal) Mesothelioma Meningioma Lung cancer (adenocarcinoma)	autosomal dominant	524,35,029	524,44,366	21874003	603089	Rahman 114 CPG
BLM	Bloom syndrome, RecQ helicase-like	BS, RECQ2, RECQL3	15q26.1	Bloom syndrome	Lymphoma and ALL hematological malignancy Myeloid hematological malignancy Squamous cell carcinoma, scc gastic, colorectal cancers	autosomal recessive	912,60,558	913,58,859	7585968	604610	Rahman 114 CPG
BMPR1A	bone morphogenetic protein receptor, type IA	ALK3, CD292	10q22.3	Juvenile polyposis syndrome	Colorectal cancer, gastric cancer, hamartoma	autosomal dominant	885,16,396	886,84,945	11381269	601299	Rahman 114 CPG
BRCA1	breast cancer 1, early onset	BRCC1, PPP1R53, RNF53	17q21	Hereditary breast-ovarian cancer	Breast cancer Ovarian cancer	autosomal dominant	411,96,312	413,22,290	7545954	113705	Rahman 114 CPG

BRCA2	breast cancer 2, early onset	FANCD1	13q12	Hereditary breast-ovarian cancer Fanconi anaemia (D1) (biallelic mutations)	Biallelic mutations: Myeloid hematological malignancy (Medulloblastoma Wilms tumor Monoallelic mutations: Breast cancer Ovarian cancer Prostate cancer Pancreas cancer	autosomal recessive autosomal dominant	328,89,611	329,73,805	8524414	600185	Rahman 114 CPG
BRIP1	BRCA1 interacting protein C-terminal helicase 1	FANCJ, BACH1	17q22	Fanconi anaemia (J) (biallelic mutations)	Biallelic mutations: Myeloid hematological malignancy Squamous cell carcinoma (head and neck, esophagus, genital tract) Monoallelic mutations: Breast cancer Ovarian cancer	autosomal recessive autosomal dominant	597,59,985	599,40,755	16153896 16116424 16116423	605882	Rahman 114 CPG
BUB1B	budding uninhibited by benzimidazoles 1 homolog beta (yeast)	BUBR1, MAD3L, SSK1	15q15	Mosaic variegated aneuploidy Syndrome	Wilms Tumor Rhabdomyosarcoma Myeloid hematological malignancy	autosomal recessive	404,53,224	405,13,337	15475955	602860	Rahman 114 CPG
CBL	Cbl proto-oncogene, E3 ubiquitin protein ligase	CBL2, RNF55, C-Cbl, FRA11B, NSLL	11q23	Noonan syndrome	JMML	autosomal dominant	1190,76,752	1191,78,859	20694012	165360	Rahman 114 CPG
CDC73	cell division cycle 73, Paf1/RNA polymerase II complex component, homolog (S. cerevisiae)	HRPT2, parafibromin	1q25	Hyperparathyroidism-jaw tumor syndrome	Parathyroid cancer Ossifying fibroma (bone)	autosomal dominant	1930,91,147	1932,23,031	12434154	607393	Rahman 114 CPG

CDH1	cadherin 1, type 1, E-cadherin (epithelial)	UVO, CD324	16q22.1	Hereditary diffuse gastric cancer	Breast cancer (lobular) Gastric cancer (diffuse)	autosomal dominant	687,71,128	688,69,445	9537325	192090	Rahman 114 CPG
CDK4	cyclin-dependent kinase 4	PSK-J3	12q14		Melanoma	autosomal dominant	581,42,034	581,49,796	8528263	123829	Rahman 114 CPG
CDKN1B	cyclin-dependent kinase inhibitor 1B (p27, Kip1)	KIP1, P27KIP1, CDKN4, MEN4, MEN1B, P27Kip1	12p.13.1		Thyroid cancer, Pituitary adenoma	autosomal recessive autosomal dominant	128,67,992	128,75,305	17030811	600778	Rahman 114 CPG
CDKN2A	cyclin-dependent kinase inhibitor 2A	p16, p14ARF	9p21		Melanoma [p16 and p14ARF] Pancreas cancer [p16] Astrocytoma [p14ARF]	autosomal dominant	219,67,751	219,95,300	7987387 7987388	600160	Rahman 114 CPG
CEBPA	CCAAT/enhancer binding protein (C/EBP), alpha	CEBP	19q13.1		Myeloid hematological malignancy	autosomal dominant	337,90,840	337,93,470	15575056	116897	Rahman 114 CPG
CHEK2	checkpoint kinase 2	CDS1, CHK2	22q12.1		Breast cancer	autosomal dominant	290,83,731	291,38,410	11967536 12094328	604373	Rahman 114 CPG
COL7A1	collagen, type VII, alpha 1	Long-chain collagen, EBD1,EBDCT, EBR1	3p21.31	Epidermolysis bullosa	Squamous cell carcinmona (skin)	autosomal recessive autosomal dominant	486,01,506	486,32,700	8513326	120120	Rahman 114 CPG
CYLD	cylindromatosis (turban tumor syndrome)	USPL2	16q12.1	Brooke-Spiegler syndrome	Cylindroma spiroadenocarcinoma Basal cell carcinoma	autosomal dominant	507,75,961	508,35,846	10835629	605018	Rahman 114 CPG
DDB2	damage-specific DNA binding protein 2, 48kDa	XPE	11p12	Xeroderma Pigmentosum (E)	Basal cell carcinoma Squamous cell carcinoma Melanoma	autosomal dominant	472,36,493	472,60,767	8798680	600811	Rahman 114 CPG
DICER1	dicer 1, ribonuclease type III	DCR1, Dicer, HERNA, MNG1	14q32.13	DICER1 syndrome	Pleuropulmonary blastoma Cystic nephroma Ovarian sex cord tumor	autosomal dominant	955,52,565	956,24,347	19556464	606241	Rahman 114 CPG

DIS3L2	DIS3 mitotic control homolog (S. cerevisiae)-like 2	FAM6A	2q37.1	Perlman syndrome	Wilms tumor	autosomal recessive	2328,25,955	2332,09,060	22306653	614184	Rahman 114 CPG
DKC1	dyskeratosis congenita 1, dyskerin	DKC, NOLA4, NAP57, XAP10, Dyskerin, CBF5, DKCX,	Xq28	Dyskeratosis congenita	acute myeloid leukemia Squamous cell carcinoma (head + neck, anorectal)	X-linked recessive	1539,91,031	1540,05,964	9590285	300126	Rahman 114 CPG
DOCK8	dedicator of cytokinesis 8	ZIR8, MRD2, FLJ00026, FLJ00152, FLJ0346	9p24.3	HyperIgE syndrome	Squamous cell carcinoma Lymphoma	autosomal recessive	2,14,865	4,65,255	19776401	611432	Rahman 114 CPG
EGFR	epidermal growth factor receptor	ERBB1	7p12		Non-small cell lung cancer	autosomal dominant	550,86,714	553,24,313	16258541	131550	Rahman 114 CPG
ELANE	elastase, neutrophil expressed	ELA2, HLE, HLN, NE, SCN, GE, PMN-E	19p13.3	Severe congenital neutropenia	Leukemia	autosomal dominant	851014	856242	11001877	130130	Rahman 114 CPG
ERCC2	excision repair cross-complementing rodent repair deficiency, complementation group 2	XPD	19q13.3	Xeroderma pigmentosum (D)	Basal cell carcinoma Squamous cell carcinoma Melanoma	autosomal recessive	458,54,246	458,73,876	7849702	126340	Rahman 114 CPG
ERCC3	excision repair cross-complementing rodent repair deficiency, complementation group 3	XPB	2q21	Xeroderma pigmentosum (B)	Basal cell carcinoma Squamous cell carcinoma Melanoma	autosomal recessive	1280,14,866	1280,51,752	2167179	133510	Rahman 114 CPG
ERCC4	excision repair cross-complementing rodent repair deficiency, complementation group 4	XPF	16p13.12	Xeroderma pigmentosum (F) Fanconi anaemia (Q)	Basal cell carcinoma Squamous cell carcinoma Melanoma	autosomal recessive	140,14,014	140,46,202	8797827	133520	Rahman 114 CPG
ERCC5	excision repair cross-complementing rodent repair deficiency, complementation group 5	XPG	13q33	Xeroderma pigmentosum (G)	Basal cell carcinoma Squamous cell carcinoma Melanoma	autosomal recessive	1034,59,705	1035,28,345	7951246	133530	Rahman 114 CPG
EXT1	exostosin 1	LGCR, LGS	8q24.11		Chondrosarcoma	autosomal dominant	1188,06,729	1191,24,092	7550340	608177	Rahman 114 CPG
EXT2	exostosin 2	SOTV	11p12-p11		Chondrosarcoma	autosomal dominant	441,17,099	442,66,979	8782816	608210	Rahman 114 CPG
FAH	fumarylacetoacetate hydrolase (fumarylacetoacetase)	FAA	15q25.1	Tyrosinemia	Hepatocellular carcinoma	autosomal recessive	804,45,122	804,79,288	8318997	276700	Rahman 114 CPG

FANCA	Fanconi anemia, complementation group A	FANCH	16q24.3	Fanconi anaemia (A)	Myeloid hematological malignancy Squamous cell carcinoma (head and neck, esophagus, genital tract)	autosomal recessive	898,03,957	898,83,065	8896564 8896563	607139	Rahman 114 CPG
FANCC	Fanconi anemia, complementation group C	FAC, FACC	9q22.3	Fanconi anaemia (C)	Myeloid hematological malignancy Squamous cell carcinoma (head and neck, esophagus, genital tract)	autosomal recessive	978,61,336	980,79,991	1574115	613899	Rahman 114 CPG
FANCG	Fanconi anemia, complementation group G	XRCC9	9p13	Fanconi anaemia (G)	Myeloid hematological malignancy Squamous cell carcinoma (head and neck, esophagus, genital tract)	autosomal recessive	350,73,832	350,80,013	9806548	602956	Rahman 114 CPG
FH	fumarate hydratase	fumarase	1q42.1	Hereditary leiomyomatosis and renal cell cancer (HLRCC)	Renal cell cancer Leiomyosarcoma (uterus)	autosomal recessive autosomal dominant	2416,60,903	2416,83,061	11865300	136850	Rahman 114 CPG
FLCN	folliculin	BHD	17p11.2	Birt-Hogg-Dube syndrome	Renal cell cancer Oncocytoma	autosomal dominant	171,15,526	171,40,502	12204536	607273	Rahman 114 CPG
GATA2	GATA binding protein 2	NFE1B	3q21.3	Emberger MonoMAC syndrome	Myeloid hematological malignancy	autosomal dominant	1281,98,270	1282,12,028	21892158 21892162 21765025 21670465	137295	Rahman 114 CPG
GBA	glucosidase, beta, acid	GLUC, GBA1, GCB	1q21	Gauchers type 1	Myeloma Lymphoma Hepatocellular carcinoma	autosomal recessive	1552,04,239	1552,14,653	2880291	606463	Rahman 114 CPG

GJB2	gap junction protein, beta 2, 26kDa	CX26, DFNA3, DFNB1, NSRD1, DFNA3A, DFNB1A, HID, KID	13q12.11	Keratosis-icthyosis-deafness syndrome (KID)	Squamous cell carcinoma	autosomal dominant	207,61,609	207,67,037	11912510	121011	Rahman 114 CPG
GPC3	glypican 3	SDYS, DGSX	Xq26.1	Simpson-Golabi-Behmel syndrome	Wilms tumor Hepatoblastoma, hepatocellular carcinoma Neuroblastoma Gonadoblastoma	X-linked recessive	1326,69,773	1331,19,922	8589713	300037	Rahman 114 CPG
HFE	hemochromatosis	HLA-H, HFE, MVDC7, HH, HLAH, TFQTL2	6p22.2	Haemochromatosis	Hepatocellular carcinoma Cholangiocarcinoma	autosomal recessive	260,87,448	260,98,571	8696333	613609	Rahman 114 CPG
HMBS	hydroxymethylbilane synthase	PBGD, UPS, PORC, PBG-D	11q23.3	Porphyria (AI)	hepatocellular carcinoma	autosomal dominant	1189,55,576	1189,64,259	2563167	609806	Rahman 114 CPG
HRAS	v-Ha-ras Harvey rat sarcoma viral oncogene homolog	HRAS1	11p15.5	Costello syndrome	Rhabdomyosarcoma Neuroblastoma Transitional cell carcinoma (bladder)	autosomal dominant	5,32,242	5,37,287	16170316	190020	Rahman 114 CPG
ITK	IL2-inducible T-cell kinase	EMT, LYK, PSCTK2,	5q33.3	Lymphoproliferative syndrome 1	Hodgkins lymphoma	autosomal recessive	1565,69,944	1566,82,201	19425169	186973	Rahman 114 CPG
KIT	v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog	PBT, C-Kit, CD117, SCFR	4q12		Gastro-Intestinal Stromal Tumor	autosomal dominant	555,24,085	556,06,881	9697690	164920	Rahman 114 CPG
MAX	MYC associated factor X	bHLHd4	14q23	Familial paraganglioma-pheochromocytoma syndrome	Paraganglioma Pheochromocytoma	autosomal dominant	654,72,892	655,69,413	21685915	154950	Rahman 114 CPG
MEN1	multiple endocrine neoplasia I	menin	11q13	Multiple endocrine neoplasia Type 1	Parathyroid, pituitary adenoma Neuroendocrine tumor Carcinoid tumor Adrenocortical carcinoma	autosomal dominant	645,70,988	645,78,766	9103196	613733	Rahman 114 CPG
MET	met proto-oncogene (hepatocyte growth factor receptor)	HGFR, c-Met	7q31		Renal cell cancer (papillary carcinoma)	autosomal dominant	1163,12,248	1164,38,440	9140397	164860	Rahman 114 CPG

MLH1	mutL homolog 1, colon cancer, nonpolyposis type 2 (E. coli)	COCA2, HNPCC2	3p21.3	MMR deficiency syndrome (biallelic mutations) Lynch syndrome / Hereditary Non-Polyposis Colon Cancer (monoallelic mutations)	Biallelic mutations: Brain tumors Hematological malignancy Embryonal tumors Monoallelic mutations: Colorectal cancer Endometrial cancer Ovarian cancer	autosomal recessive autosomal dominant	370,34,823	371,07,380	8128251 8145827	120436	Rahman 114 CPG
MSH2	mutS homolog 2, colon cancer, nonpolyposis type 1 (E. coli)	COCA1, HNPCC1	2p21	MMR deficiency syndrome (biallelic mutations) Lynch syndrome / Hereditary Non-Polyposis Colon Cancer (monoallelic mutations)	Biallelic mutations: Brain tumors Hematological malignancy Embryonal tumors Monoallelic mutations: Colorectal cancer Endometrial cancer Ovarian cancer Sebaceous adenoma, carcinoma, epithelioma	autosomal recessive autosomal dominant	476,30,108	477,89,450	8252616 8261515	609309	Rahman 114 CPG
MSH6	mutS homolog 6 (E. coli)	GTBP	2p16	MMR deficiency syndrome (biallelic mutations) Lynch syndrome / Hereditary Non-Polyposis Colon Cancer (monoallelic mutations)	Biallelic mutations: Brain tumors Hematological malignancy Embryonal tumors Monoallelic mutations: Colorectal cancer Endometrial cancer Ovarian cancer	autosomal recessive autosomal dominant	480,10,221	480,34,092	9354786	600678	Rahman 114 CPG

MTAP	methylthioadenosine phosphorylase	MSAP, C86fus, BDMF, DMSFH, DMSMFH, LGMBF	9p21.3	Diaphyseal medullary stenosis with malignant fibrous histiocytoma (DMS-MFH)	malignant fibrous histiocytoma (sarcoma)	autosomal dominant	218,02,542	219,31,646	22464254	156540	Rahman 114 CPG
MUTYH	mutY homolog (E. coli)	MYH	1p34.1		Colorectal cancer	autosomal recessive	457,94,835	458,06,142	11818965	604933	Rahman 114 CPG
NBN	nibrin	NBS1	8q21	Nijmegen breakage syndrome	Lymphoma Medulloblastoma Glioma Rhabdomyosarcoma	autosomal recessive	909,45,564	910,15,456	9590180 9620777	602667	Rahman 114 CPG
NF1	neurofibromin 1	NFNS, VRNF, WSS	17q12	Neurofibromatosis type 1	Glioma Malignant peripheral nerve sheath tumor	autosomal dominant	294,21,945	297,08,905	2134734 1694727	613113	Rahman 114 CPG
NF2	neurofibromin 2 (merlin)	ACN, BANF, SCH	22q12.2	Neurofibromatosis type 2	Vestibular schwannoma Meningioma Ependymoma	autosomal dominant	299,99,545	300,94,587	8453669 8379998	607379	Rahman 114 CPG
PALB2	partner and localizer of BRCA2	FANCN	16p12.1	Fanconi anaemia (N) (biallelic mutations)	Biallelic mutations: Myeloid hematological malignancy Medulloblastoma Neuroblastoma Wilms tumor Monoallelic mutations: Breast cancer Pancreas cancer	autosomal recessive autosomal dominant	236,14,488	236,52,631	17200671 17200672 17200668 17287723	610355	Rahman 114 CPG
PDGFRA	platelet-derived growth factor receptor, alpha polypeptide	PDGFR2, RHEPDGFRA, CD140a, CD140A, PDGFRA/BCR Fusion	4q12		Gastro-Intestinal Stromal Tumor	autosomal dominant	550,95,264	551,64,414	14699510	173490	Rahman 114 CPG
PHOX2B	paired-like homeobox 2b	PMX2B	4p12		Neuroblastoma	autosomal dominant	417,46,099	417,50,987	12640453	603851	Rahman 114 CPG

PMS2	PMS2 postmeiotic segregation increased 2 (S. cerevisiae)	PMSL2, HNPCC4	7p22	MMR deficiency syndrome (biallelic mutations) Lynch syndrome / Hereditary Non-Polyposis Colon Cancer (monoallelic mutations)	Biallelic mutations: Brain tumors Hematological malignancy Supratentorial primitive neuroectodermal tumors Monoallelic mutations: Colorectal cancer Endometrial cancer Ovarian cancer	autosomal recessive autosomal dominant	60,12,870	60,48,756	8072530	600259	Rahman 114 CPG
POLD1	polymerase (DNA directed), delta 1, catalytic subunit	POLD, CDC2, CRCS 10, MDPL	19q13.33	PPAP (polymerase proofreading associated polyposis)	Colorectal cancer Endometrial cancer	autosomal dominant	508,87,461	509,21,273	23263490	174761	Rahman 114 CPG
POLE	polymerase (DNA directed), epsilon, catalytic subunit	FILS, POLE1, CRCS12	12q24.33	PPAP (polymerase proofreading associated polyposis)	Colorectal cancer	autosomal recessive autosomal dominant	1332,00,348	1332,63,951	23263490	174762	Rahman 114 CPG
POLH	polymerase (DNA directed), eta	XPV, XP-V, RAD30, RAD30A	6p21.1	Xeroderma pigmentosa V	Squamous cell cancer (skin)	autosomal recessive	435,43,887	435,86,701	10385124	603968	Rahman 114 CPG
PRKAR1A	protein kinase, cAMP-dependent, regulatory, type I, alpha	TSE1	17q23-q24	Carney complex	Myxoma (cardiac/cutaneous /breast) Thyroid cancer Sex cord-stromal tumor	autosomal dominant	665,08,110	665,28,908	10973256	188830	Rahman 114 CPG
PRSS1	protease, serine, 1 (trypsin 1)	TRP1, TRY1, TRY4, TRYP1	7q34		Pancreatic cancer	autosomal dominant	1424,57,319	1424,60,923	8841182	276000	Rahman 114 CPG
PTCH1	patched 1	PTCH, NBCCS, BCNS	9q22.3	Nevoid basal cell carcinoma syndrome Gorlin Syndrome	Basal cell carcinoma Medulloblastoma	autosomal dominant	982,05,262	982,79,339	8658145 8681379	601309	Rahman 114 CPG

PTEN	phosphatase and tensin homolog	BZN, MHAM	10q23.3	Cowden Syndrome PTEN hamartoma tumor syndrome	Breast cancer Thyroid cancer Endometrial cancer	autosomal dominant	896,22,870	897,31,687	9140396	601728	Rahman 114 CPG
PTPN11	protein tyrosine phosphatase, non-receptor type 11	CFC, NS1, SHP2, BPTP3, PTP2C, PTP-1D, SH-PTP2, SH-PTP3	12q24.13	Noonan syndrome	JMML neuroblastoma	autosomal dominant	1128,56,155	1129,47,717	11704759 12717436	176876	Rahman 114 CPG
RAD51C	RAD51 homolog C (S. cerevisiae)	RAD51L2	17q25.1	Fanconi anaemia (O) (biallelic mutations)	Monoallelic mutations: Ovarian cancer	autosomal recessive autosomal dominant	567,69,934	568,11,703	20400964	602774	Rahman 114 CPG
RAD51D	RAD51 homolog D (S. cerevisiae)	RAD51L3	17q11		Ovarian cancer	autosomal dominant	334,26,811	334,48,541	21822267	602954	Rahman 114 CPG
RB1	retinoblastoma 1	OSRC, RB	13q14.2		Retinoblastoma Pinealoma Sarcoma Melanoma	autosomal dominant	488,77,883	490,56,122	2885916	614041	Rahman 114 CPG
RECQL4	RecQ protein-like 4	RecQ4	8q24.3	Rothmund-Thompson syndrome	Osteosarcoma Basal cell carcinoma Squamous cell carcinoma	autosomal recessive	1457,36,667	1457,43,229	10319867	603780	Rahman 114 CPG
RET	ret proto-oncogene	HSCR1	10q11.2	Multiple endocrine neoplasia 2A/2B Familial medullary thyroid carcinoma	Medullary thyroid cancer Pheochromocytoma	autosomal dominant	435,72,475	436,25,799	8099202	164761	Rahman 114 CPG
RHBDF2	rhomboid 5 homolog 2 (Drosophila)	RHBDL6	17q25.1		Esophageal cancer	autosomal dominant	744,66,976	744,97,508	22265016	614404	Rahman 114 CPG
RMRP	RNA component of mitochondrial RNA processing endoribonuclease	CHH, RMRPR	9p13.3	Cartilage-hair hypoplasia syndrome	Non-hodgkin lymphoma Squamous carcinoma (bcc) Leukemia	autosomal recessive	35657748	356,58,015	11207361	157660	Rahman 114 CPG
RUNX1	runt-related transcription factor 1	AML1, CBFA2	21q22.3		Myeloid hematological malignancy (leukemia)	autosomal dominant	361,60,098	373,57,047	10508512	151385	Rahman 114 CPG
SBDS	Shwachman-Bodian-Diamond syndrome	SDS	7q11	Schwachman-Diamond syndrome	Myeloid hematological malignancy	autosomal recessive	664,52,664	664,60,635	12496757	607444	Rahman 114 CPG

SDHA	succinate dehydrogenase complex, subunit A, flavoprotein (Fp)	FP, PGL5, SDH1, SDH2, SDHF, CMD1GG	5p15.33	Carney-Stratakis syndrome	Paraganglioma Pheochromocytoma Gastrointestinal stromal tumor (GIST)	autosomal recessive autosomal dominant	2,18,356	2,56,815	20484225	600857	Rahman 114 CPG
SDHAF2	succinate dehydrogenase complex assembly factor 2	SDH5, PGL2	11q12.2	Familial paraganglioma-pheochromocytoma syndrome	Paraganglioma Pheochromocytoma	autosomal dominant	611,97,514	612,15,001	19628817	613019	Rahman 114 CPG
SDHB	succinate dehydrogenase complex, subunit B, iron sulfur (Ip)	SDH1	1p36.1-p35	Familial paraganglioma-pheochromocytoma syndrome	Paraganglioma Pheochromocytoma Renal cell cancer	autosomal dominant	173,45,217	173,80,665	11404820	185470	Rahman 114 CPG
SDHC	succinate dehydrogenase complex, subunit C, integral membrane protein, 15kDa	PGL3	1q21	Familial paraganglioma-pheochromocytoma syndrome	Paraganglioma Pheochromocytoma Gastrointestinal stromal tumor (GIST)	autosomal dominant	1612,84,047	1613,32,984	11062460	602413	Rahman 114 CPG
SDHD	succinate dehydrogenase complex, subunit D, integral membrane protein	PGL1	11q23	Familial paraganglioma-pheochromocytoma syndrome	Paraganglioma Pheochromocytoma Gastrointestinal stromal tumor (GIST)	autosomal dominant	1119,57,497	1119,90,353	10657297	602690	Rahman 114 CPG
SERPINA1	serpin peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 1	PRO0684, A1A, A1AT, AAT, PI, PI1, PRO2275, alpha1AT	14q32.13	Alpha1 antitrypsin defiency	Hepatocellular carcinoma	autosomal recessive	948,43,084	948,57,030	3485248 7045697	107400	Rahman 114 CPG
SH2D1A	SH2 domain containing 1A	RP5-1052M9.3, DSHP, EBVS, IMD5, LYP, MTCP1, SAP, SAP/SH2D1A, XLP, XLPD	Xq25	Lymphoproliferative disease	Lymphoma	X-linked recessive	1234,80,194	1235,07,005	9771704	300490	Rahman 114 CPG
SLC25A13	solute carrier family 25 (aspartate/glutamate carrier), member 13	ARALAR2, CITRIN, CTLN2	7q21.3	Citrullinaemia	Hepatocellular carcinoma	autosomal recessive	957,49,532	959,51,459	10369257	603859	Rahman 114 CPG

SMAD4	SMAD family member 4	MADH4, DPC4	18q21.1	Juvenile polyposis syndrome	Colorectal cancer	autosomal dominant	484,94,410	486,11,415	9582123	600993	Rahman 114 CPG
SMARCA4	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 4	BAF190, BAF190A, BRG1, MRD16, RTPS2, SNF2, SNF2L4, SNF2LB, SWI2, hSNF2b	19p13.2	Rhabdoid predisposition syndrome	Rhabdoid tumor	autosomal dominant	110,71,598	111,76,071	20137775	603254	Rahman 114 CPG
SMARCB1	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily b, member 1	INI1, SNF5L1	22q11	Rhabdoid predisposition syndrome	Rhabdoid tumor (renal, extra-renal) Central primitive neuroectodermal tumor	autosomal dominant	241,29,150	241,76,703	10521299	601607	Rahman 114 CPG
SMARCE1	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily e, member 1	BAF57	17q21.2		Mengingioma	autosomal dominant	387,81,214	388,04,760	23377182	603111	Rahman 114 CPG
SOS1	son of sevenless homolog 1 (Drosophila)	GF1, GGF1, GINGF, HGF, NS4	2p22.1	Noonan syndrome	Rhabdomyosarcoma	autosomal dominant	392,08,537	393,51,486	17143285	182530	Rahman 114 CPG
SRY	sex determining region Y	TDF, TDY, SRXX1, SRXY1	Yp11.31		Gonadoblastoma	Y-linked	26,54,896	26,55,740	2247149 2247151	480000	Rahman 114 CPG
STAT3	signal transducer and activator of transcription 3 (acute-phase response factor)	APRF, HIES	17q21.1	Hyper-immunoglobulin E syndrome	Lymphoma	autosomal dominant	404,65,342	405,40,586	17676033	102582	Rahman 114 CPG
STK11	serine/threonine kinase 11	LKB1	19p13.3	Peutz-Jeghers syndrome	Colorectal cancer Gastric cancer Breast cancer Sex cord-stromal tumor	autosomal dominant	12,05,798	12,28,434	9425897 9428765	602216	Rahman 114 CPG
SUFU	suppressor of fused homolog (Drosophila)	SUFUH, SUFUXL	10q24.32		Medulloblastoma, meningioma	autosomal dominant	1042,63,744	1043,93,292	12068298	607035	Rahman 114 CPG

TERT	telomerase reverse transcriptase	CMM9, DKCA2, DKCB4, EST2, PFBMFT1, TCS1, TP2, TRT, hEST2, hTRT	5p15.33	Dyskeratosis congenita	acute myeloid leukemia Squamous cell carcinoma (head + neck, anorectal) Melanoma	autosomal recessive autosomal dominant	12,53,262	12,95,184	16247010	187270	Rahman 114 CPG
TGFBR1	transforming growth factor, beta receptor 1	RP11-96L7.1, AAT5, ACVRLK4, ALK-5, ALK5, LDS1A, LDS2A, MSSE, SKR4, TGFR-1	9q22.33	Multiple self-healing squamous epithelioma (MSSE) Ferguson-Smith syndrome	Squamous cell carcinoma (skin)	autosomal dominant	1018,66,320	1019,16,474	21358634	190181	Rahman 114 CPG
TMEM127	transmembrane protein 127		2q11.2		Pheochromocytoma	autosomal dominant	969,16,312	969,31,732	20154675	613403	Rahman 114 CPG
TNFRSF6	transforming growth factor, beta receptor 1	FAS, RP11-96L7.1, AAT5, ACVRLK4, ALK-5, ALK5, LDS1A, LDS2A, MSSE, SKR4, TGFR-1	10q23.31	Autoimmunie lymphoproliferative syndrome	Lymphoma	autosomal dominant	907,50,414	907,75,542	7540117	134637	Rahman 114 CPG
TP53	tumor protein p53	LFS1	17p13.1	Li-Fraumeni syndrome	Breast cancer Sarcoma Adrenocortical carcinoma Astrocytoma	autosomal dominant	75,65,097	75,90,856	1978757	191170	Rahman 114 CPG
TRIM37	tripartite motif containing 37	MUL, POB1, TEF3	17q22	Mulibrey-nanism	Wilms tumor	autosomal recessive	570,59,999	571,84,282	10888877	605073	Rahman 114 CPG
TSC1	tuberous sclerosis 1	LAM, hamartin	9q34	Tuberous sclerosis 1	Renal cell cancer, angiomyolipoma Subependymal giant cell astrocytoma Rhabdomyoma (cardiac)	autosomal dominant	1357,66,735	1358,20,020	9242607	605284	Rahman 114 CPG

TSC2	tuberous sclerosis 2	TSC4, tuberin	16p13.3	Tuberous sclerosis 2	Renal cell cancer, angiomyolipoma Subependymal giant cell astrocytoma Rhabdomyoma (cardiac)	autosomal dominant	20,97,466	21,38,716	8269512	191092	Rahman 114 CPG
UROD	uroporphyrinogen decarboxylase	RP11-69J16.2, PCT, UPD	1p34.1	Porphyria (cutanea tarda)	hepatocellular carcinoma	autosomal recessive autosomal dominant	454,77,819	454,81,247	3775362	613521	Rahman 114 CPG
VHL	von Hippel-Lindau tumor suppressor, E3 ubiquitin protein ligase	VHL1	3p25	Von Hippel-Lindau syndrome	Renal cell cancer Pheochromocytoma Neuroendocrine tumor (pancreas) Hemangioblastoma (central nervous system, retina)	autosomal dominant	101,82,692	101,93,904	8493574	608537	Rahman 114 CPG
WAS	Wiskott-Aldrich syndrome	IMD2, SCNX, THC, THC1, WASP	Xp11.23	Wiskott-Aldrich syndrome WAS-related syndrome	Lymphoma	X-linked recessive	485,34,985	485,49,818	8069912	300392	Rahman 114 CPG
WRN	Werner syndrome, RecQ helicase-like	RECQ3, RECQL2	8p12	Werner syndrome	Sarcoma Melanoma Thyroid cancer	autosomal recessive	308,91,317	310,31,285	8602509	604611	Rahman 114 CPG
WT1	Wilms tumor 1	AWT1, EWS-WT1, GUD, NPHS4, WAGR, WIT-2, WT33	11p13	WAGR syndrome Denys-Drash syndrome Frasier syndrome	Wilms tumor Gonadoblastoma	autosomal dominant	324,09,321	324,57,176	1673293	607102	Rahman 114 CPG
XPA	xeroderma pigmentosum, complementation group A	XP1, XPAC	9q22.3	Xeroderma pigmentosum (A)	Basal cell carcinoma Squamous cell carcinoma Melanoma	autosomal recessive	1004,37,191	1004,59,639	2234061	611153	Rahman 114 CPG
XPC	xeroderma pigmentosum, complementation group C	RAD4, XPCC	3p25	Xeroderma pigmentosum (C)	Basal cell carcinoma Squamous cell carcinoma Melanoma	autosomal recessive	141,86,647	142,20,283	8298653	613208	Rahman 114 CPG
CTR9	PROTEIN 1	SH2BP1	11p15.4		Wilm's tumor	autosomal dominant	107,50,986	107,79,754	25099282	609366	Reference (see PMID)

ETV6	ETS VARIANT GENE 6		12p13.2	familial thrombocytopenia and hematologic malignancy	pediatric ALL	autosomal dominant	116,49,853	11,895,401	25807284; 26102509; 26522332	600618	Reference (see PMID)
FANCM	FANCM GENE		14q21.2		triple negative breast cancer	autosomal dominant	451,35,932	452,00,889	25288723	609644	Reference (see PMID)
JMJD1C	JUMONJI DOMAIN-CONTAINING PROTEIN 1C		10q21.3		intracranial germ cell tumors	autosomal dominant		631,67,221	24896186	604503	Reference (see PMID)
MITF	microphthalmia-associated transcription factor		3p13		familial melanoma	autosomal dominant	697,39,434	699,68,336	22080950; 22080950	156845	Reference (see PMID)
NTHL1	ENDONUCLEASE III-LIKE 1	NTH1, OCTS3	16p13.3		colon adenocarcinoma, bladder carcinoma, meningioma, basal cell carcinoma, colorectal adenomas, head and neck squamous cell carcinoma-based on one woman case report	autosomal recessive	20,39,814	2,047,865	26559593	602656	Reference (see PMID)
PAX5	PAIRED BOX GENE 5		9p13.2		B-ALL	autosomal dominant	368,33,274	370,35,318	24013638	167414	Reference (see PMID)
POT1	PROTECTION OF TELOMERES 1		7q31.33		melanoma	autosomal dominant	1248,22,385	1249,29,982	24686849	606478	Reference (see PMID)
PRDM9	PR DOMAIN-CONTAINING PROTEIN 9		5p14.2		childhood leukemia (B-ALL)	autosomal dominant	235,07,614	235,28,596	23222848; 24754746	609760	Reference (see PMID)
RECQL	RECQ PROTEIN-LIKE		12p12.1		breast cancer	autosomal dominant	214,68,909	215,01,668	25915596	600537	Reference (see PMID)
SPRTN	SprT-LIKE N-TERMINAL DOMAIN PROTEIN		1q42.2		hepatocellular carcinoma	autosomal recessive	2313,37,935	231,370,550	25261934	616086	Reference (see PMID)

ERCC1	EXCISION REPAIR, COMPLEMENTING DEFECTIVE, IN CHINESE HAMSTER, 1	UV20	19q13.32	Cerebrooculofacioskeletal syndrome 4		autosomal recessive	454,07,332	454,78,819	26580448	126380	St. Jude 29 list NEJM paper
FANCI	FANCI GENE		15q26.1	Fanconi anemia, complementation group I		autosomal recessive	892,43,947	893,17,130	26580448; 28687356; 28961279	611360	St. Jude 29 list NEJM paper
FANCL	FANCL GENE	PHF9; FAAP43	2p16.1	Fanconi anemia, complementation group L		autosomal recessive	581,59,242	58,241,680	26580448; 29335925; 28678401	608111	St. Jude 29 list NEJM paper
NHP2	NUCLEOLAR PROTEIN FAMILY A, MEMBER 2		5q35.3	Dyskeratosis congenita, autosomal recessive 2		autosomal recessive	1781,49,462	1781,53,959	26580448; 21209122	606470	St. Jude 29 list NEJM paper
NOP10	NUCLEOLAR PROTEIN FAMILY A, MEMBER 3		15q14	Dyskeratosis congenita, autosomal recessive 1		autosomal recessive	343,41,715	34,343,160	26580448; 21209122	606471	St. Jude 29 list NEJM paper
SH2B3	SH2B ADAPTOR PROTEIN 3		12q24.12			autosomal recessive	1114,05,107	1114,51,623	26580448; 29212164	605093	St. Jude 29 list NEJM paper
BRAF	V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1	BRAF1, RAFB1	7q34	RASopathy (Cardiofaciocutaneous Syndrome, Noonan Syndrome 7, and LEOPARD Syndrome 3)		autosomal dominant	1407,15,950	1409,24,763	26580448	164757	St. Jude 60 Figure 2 NEJM paper
CDKN1C	CYCLIN-DEPENDENT KINASE INHIBITOR 1C		11p15.4	Beckwith-Wiedemann Syndrome	embryonal tumors	autosomal dominant	28,83,217	2,885,815	26580448; 10424811	600856	St. Jude 60 Figure 2 NEJM paper
EPCAM	EPITHELIAL CELLULAR ADHESION MOLECULE	TACSTD1, M4S1, GA733-2	2p21	Colorectal cancer, hereditary nonpolyposis, type 8	Hereditary Nonpolyposis Colorectal Cancer	autosomal dominant	473,69,147	473,87,027	26580448; 29237405	185535	St. Jude 60 Figure 2 NEJM paper

KRAS	V-KI-RAS2 KIRSTEN RAT SARCOMA VIRAL ONCOGENE HOMOLOG		12p12.1	RASopathy (Cardiofaciocutaneous syndrome 2, Noonan syndrome 3, RAS-associated autoimmune leukoproliferative disorder, Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic)		autosomal dominant	252,04,788	252,50,930	26580448; 25808193	190070	St. Jude 60 Figure 2 NEJM paper
MAP2K1	MITOGEN-ACTIVATED PROTEIN KINASE KINASE 1	PRKMK1, MKK1, MAPKK1, MEK1	15q22.31	RASopathy (Cardiofaciocutaneous syndrome 3)		autosomal dominant	663,86,872	664,91,543	26580448; 25899310	176872	St. Jude 60 Figure 2 NEJM paper
MAP2K2	MITOGEN-ACTIVATED PROTEIN KINASE KINASE 2	PRKMK2, MKK2, MAPKK2, MEK2	19p13.3	RASopathy (Cardiofaciocutaneous syndrome 4)		autosomal dominant	40,90,320	41,24,183	26580448; MAP2K2	601263	St. Jude 60 Figure 2 NEJM paper
NRAS	NEUROBLASTOMA RAS VIRAL ONCOGENE HOMOLOG	NRAS1	1p13.2	RASopathy (RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic; Epidermal nevus, somatic; Melanocytic nevus syndrome, congenital, somatic; Neurocutaneous melanosis, somatic; Noonan syndrome 6; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic)		autosomal dominant	1147,04,463	1147,16,893	26580448; 28594414	164790	St. Jude 60 Figure 2 NEJM paper
RAF1	V-RAF-1 MURINE LEUKEMIA VIRAL ONCOGENE HOMOLOG 1	CRAF	3p25.2	RASopathy (LEOPARD syndrome 2, Noonan syndrome 5)		autosomal dominant	125,83,600	12,664,200	26580448; 27038324	164760	St. Jude 60 Figure 2 NEJM paper
SHOC2	SUPPRESSOR OF CLEAR, C. ELEGANS, HOMOLOG OF	SUR8	10q25.2	RASopathy (Noonan-like syndrome with loose anagen hair)		autosomal dominant	1109,19,542	111,013,666	26580448; 27466182	602775	St. Jude 60 Figure 2 NEJM paper