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Search Strategies for “A comprehensive analysis of mutations in cutaneous squamous cell carcinoma reveals novel genes and mutations associated with patient-specific characteristics and metastasis”

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Search Strategies for “A comprehensive analysis of mutations in cutaneous squamous cell carcinoma reveals novel genes and mutations associated with patient-specific characteristics and metastasis”

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7/9/2019

ID Search

#1 (((akt3 OR "akt 3" OR "akt serine/threonine kinase 3" OR mpph OR mpph2 OR "pkb gamma" OR "pkb γ" OR pkbγ OR pkbγ OR prkbg OR "rac pk gamma" OR "rac pk γ" OR "rac pky" OR "racpky" OR "racpk γ" OR "rac γ" OR "racy" OR "stk 2" OR "v akt" OR "hgnc 393" OR alk OR cd246 OR "cd 246" OR nblst3 OR "nblst 3" OR apc OR "adenomatous polyposis coli" OR "deleted in polyposis 2.5" OR dp2.5 OR "dp 2.5" OR arid1a OR "arid 1a" OR eld OR "b 120" OR css2 OR "css 2" OR osa1 OR "osa 1" OR "p 270" OR bm029 OR "bm 029" OR mrd14 OR "mrd 14" OR hosa1 OR "hosa 1" OR baf250 OR "baf 250" OR "baf 250a" OR smarcf1 OR "smarcf 1" OR baf250a OR "chromosome 1 open reading frame 4" OR c1orf4 OR "b120 gene" OR b120 OR p270 OR bap1 OR "bap 1" OR uchl2 OR "uchl 2" OR hucep6 OR "hucep 6" OR hucep13 OR "hucep 13" OR cdh1 OR "cdh 1" OR cadherin OR uvomorulin OR "calcium dependent adhesion protein" OR "epithelial liver cell adhesion molecule" OR uvo OR cdhe OR ecad OR lcam OR "arc 1" OR arc1 OR "bcds 1" OR bcds1 OR "cd 324" OR cd324 OR "cdk 4" OR cdk4 OR "cyclin dependent kinase 4" OR "cmm 3" OR cmm3) NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion)))):ti,ab,kw

#2 ((("psk j3" OR pskj3 OR "pskj 3" OR "cell division kinase 4" OR cdkn2a OR "cdkn 2a" OR "cyclin dependent kinase inhibitor 2a" OR "cdk4 inhibitor" OR "multiple tumor suppressor 1" OR arf OR mlm OR p14 OR "p 14" OR p16 OR "p 16" OR p19 OR "p 19" OR cmm2 OR "cmm 2" OR ink4 OR "ink 4" OR mts1 OR "mts 1" OR tp16 OR "tp 16" OR cdk4i OR "cdk 4i" OR cdkn2 OR "cdkn 2" OR ink4a OR "ink 4a" OR p14arf OR "p14 arf" OR p19arf OR "p19 arf" OR p16ink4 OR "p16 ink4" OR p16ink4a OR "p16 ink4a" OR p16?ink4* OR p16?ink4a OR ddr2 OR "discoidin domain receptor tyrosine kinase 2" OR wrkn OR mig20a OR "mig 20a" OR tyro10 OR "tyro 10" OR "neurotrophic tyrosine kinase receptor related 3" OR ntrkr3 OR "ntrkr 3" OR "tyrosine kinase receptor related to neurotrophic trk" OR tkt OR egfr OR "epidermal growth factor receptor" OR "verb b" OR "verbb" OR "erb b" OR erbb OR erbb1 OR "erbb 1" OR her1 OR "her 1" OR "species antigen 7" OR sa7 OR "sa 7" OR mena OR pig61 OR "pig 61" OR nisbd2 OR "nisbd 2" OR "erbb 2" OR neu OR her2* OR "her 2*" OR tkr1 OR "tkr 1" OR cd340 OR "cd 340" OR "mln 19" OR mln19 OR "erb b2" OR erbb2 OR "avian erythroblastic leukemia viral oncogene" OR "neuroblastoma or glioblastoma derived oncogene" OR ngl OR erbb3* OR "erbb b3" OR "avian erythroblastic leukemia viral oncogene homolog 3" OR her3 OR "her 3" OR ferlk OR lccs2 OR "lccs 2" OR erbb3 OR "mda bf 1" OR "mda bf1" OR "mdabf 1" OR mdabf1 OR "p180 erbb3" OR p180erbb3 OR "p45 serbb3" OR p45serbb3 OR "p85 serbb3" OR p85serbb3 OR "erb b4" OR "erbb 4" OR her4 OR "her 4" OR als19 OR "als 19" OR p180erbb4 OR "p180 erbb4" OR erbb4 OR esr1 OR "esr 1" OR esra OR estrr OR nr3a1 OR "estrogen receptor 1" OR "estrogen receptor alpha" OR "fat tumor suppressor") NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion)))):ti,ab,kw

#3 (((fat1 OR "fat 1" OR me5 OR "me 5" OR cdhf7 OR "cd hf7" OR "cdhf 7" OR cdhr8 OR "cd hr8" OR "cdhr 8" OR hfat1 OR "hfat 1" OR "f box" OR "wd40 domain protein 7" OR fbxw7 OR "fbxw 7" OR fbw7 OR "fbw 7" OR fbxw6 OR "fbxw 6" OR fbw6 OR "fbw 6" OR fbxo30 OR "fbxo 30" OR fbx30 OR "fbx 30" OR "homolog of archipelago" OR cdc4 OR "cdc 4" OR "homolog of sel10" OR "homolog of sel 10" OR fgfr2 OR "fgfr 2" OR "fibroblast growth factor receptor 2" OR "fgf receptor" OR "receptor like protein tyrosine kinase 14" OR bek OR jws OR bbds OR cek3 OR "cek 3" OR cfd1 OR "cfd 1" OR ect1 OR "ect 1" OR kgfr OR tk14 OR "tk 14" OR tk25 OR "tk 25" OR "bfr 1" OR bfr1 OR cd332 OR "cd 332" OR "k sam" OR ksam OR gata3 OR "gata 3" OR "gata binding protein 3" OR hdr OR hds OR "guanine nucleotide binding protein" OR "q polypeptide" OR gnaq OR "g protein" OR "gq class" OR "g α q" OR gaq OR gaq OR sws OR cmc1 OR "cmc 1" OR "g alpha q" OR scg6 OR "scg 6" OR sgvi OR "sg vi" OR "gnas 1" OR pita3 OR "pita 3" OR c20orf45 OR gnas OR gnas1 OR aho OR gsa OR gsp OR poh OR gpsa OR nesp OR hnf1a OR "hnf 1a" OR lfb1 OR "lfb 1" OR tcf1 OR "tcf 1" OR hnf4a OR "hnf 4a" OR mody3 OR "mody 3" OR iddm20 OR "hnf1 homeobox a" OR "transcription factor 1" OR "hepatocyte nuclear factor 1 alpha" OR "hepatocyte nuclear factor 1α" OR "hepatocyte nuclear factor 1 α" OR "hepatocyte nuclear factor 1" OR hnf1 OR "hnf 1" OR "albumin proximal factor" OR hras OR hras1 OR "hras 1" OR rash1 OR "rash 1" OR p21?ras* OR p21 OR ctlo OR kras OR hamsv OR kras2 OR "kras 2" OR rask2 OR "rask 2" OR "ki ras" OR "c bas/has" OR p21ras OR "c h ras" OR chras OR "c bas?has" OR "c k ras" OR ckras OR "h rasidx" OR "c ki ras" OR ckiras OR "c ha ras1") NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR

missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ti,ab,kw

#4 (((idh1 OR "idh 1" OR "isocitrate dehydrogenase 1" OR "peroxisomal isocitrate dehydrogenase" OR picd OR idpc OR icdc OR idh OR idp OR idcd OR "hel 216" OR hel216 OR "hel s 26" OR "hels 26" OR hels26 OR "hel s26" OR idh2 OR "idh 2" OR "isocitrate dehydrogenase 2" OR idpm OR idhm OR "icd m" OR d2hga2 OR "d2 hga2" OR "d2hga 2" OR jak2 OR "jak 2" OR jtk10 OR "jtk 10" OR thcyt3 OR "thcyt 3" OR "janus kinase 2" OR kdr OR "kinase insert domain receptor" OR "tyrosine kinase growth factor receptor" OR flk1 OR "flk 1" OR "vascular endothelial growth factor receptor" OR vegfr2 OR cd309 OR "cd 309" OR vegfr OR keep1 OR "keep 1" OR "kelch like ech associated protein 1" OR inrf2 OR "inrf 2" OR klhl19 OR "klhl 19" OR kit OR "mast cell growth factor receptor" OR "stem cell factor receptor" OR scfr OR fdc OR ssm OR sco1 OR "sco 1" OR sco5 OR "sco 5" OR sow3 OR "sow 3" OR cd117 OR "cd 117" OR "c kit" OR "tr kit" OR gsfsc1 OR "gsfsc 1" OR gsfsc5 OR "gsfsc 5" OR gsfscow3 OR "gsfscow 3" OR "lysine specific methyltransferase 2c" OR halr OR ml13 OR "ml 3" OR klefs2 OR "klefs 2" OR kmt2c OR "kmt 2c" OR "lysine methyltransferase 2c" OR kiaa1506 OR "kiaa 1506" OR kms OR ml12 OR "ml 2" OR ml14 OR "ml 4" OR aad10 OR "aad 10" OR kabuk1 OR "kabuk 1" OR tnrc21 OR "tnrc 21" OR cag114 OR "cag 114" OR kmt2d OR "kmt 2d" OR "lysine methyltransferase 2d" OR "all1 related" OR "all 1 related" OR alr OR "mitogen activated protein kinase" OR "mapk?erk kinase 1" OR mek1 OR "mek 1" OR cfc3 OR "cfc 3" OR mkk1 OR "mkk 1" OR mapkk1 OR "mapkk 1" OR prkmk1 OR "prkmk 1" OR map2k1 OR "mitogen activated protein kinase kinase 1" OR cfc4 OR "cfc 4" OR mkk2 OR "mkk 2" OR mapkk2 OR "mapkk 2" OR prkmk2 OR "prkmk 2" OR map2k2 OR "mitogen activated protein kinase kinase 2" OR "mapk?erk kinase 2" OR mek2 OR "mek 2" OR "mitogen activated kinase kinase kinase 1" OR map3k1 OR "map?erk kinase kinase 1" OR mekk1 OR "mekk 1" OR mapkkk1 OR "mapkkk 1" OR "mek kinase" OR mekk) NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ti,ab,kw

#5 (((srxy6 OR "srxy 6" OR auts9 OR "auts 9" OR rccp2 OR "rccp 2" OR "c met" OR dfnb97 OR "dfnb 97" OR "met proto oncogene" OR "met oncogene" OR "hepatocyte growth factor receptor" OR hgfr OR notch1 OR "notch 1" OR "notch receptor 1" OR "translocation associated notch homolog" OR tan1 OR "tan 1" OR hn1 OR "hn 1" OR aos5 OR "aos 5" OR aovd1 OR "aovd 1" OR ns6 OR "ns 6" OR cmns OR ncms OR alps4 OR "alps 4" OR "n ras" OR nras1 OR "nras 1" OR nras OR pdgfra OR "pdgfr α " OR cd140a OR "cd 140a" OR pdgfra OR "platelet derived growth factor receptor alpha" OR "platelet derived growth factor receptor α " OR pdgfr2 OR "pdgfr 2" OR "phosphatidylinositol 3 kinase" OR pik3ca OR "p110 alpha" OR "p110 α " OR p110 α OR "pi3k alpha" OR "pi3k α " OR pi3k α OR "pik3 alpha" OR "pik3 α " OR pik3 α OR mcm OR cws5 OR "cws 5" OR mcap OR pi3k OR clapo OR clove OR mcmtc OR "phosphatidylinositol 4,5 biphosphate 3 kinase catalytic subunit alpha" OR "phosphatidylinositol 4,5 biphosphate 3 kinase catalytic subunit α " OR pten OR bzs OR dec OR cws1 OR "cws 1" OR glm2 OR "glm 2" OR mham OR tep1 OR "tep 1" OR mmac1 OR "mmac 1" OR pten1 OR "pten 1" OR 10q23del OR ptenbeta OR pten β OR "pten phosphatase and tensin homolog" OR "mutated in multiple advanced cancers 1" OR "phosphatase and tensin homolog deleted on chromosome 10" OR rb1 OR "rb 1" OR "rb transcriptional corepressor 1" OR rb OR prb OR oscr OR pp110 OR "pp 110" OR ppp1r130 OR "ppp1r 130" OR "p105 rb" OR ret OR ptc OR mtc1 OR "mtc 1" OR hscr1 OR "hscr 1" OR men2a OR "men 2a" OR

men2b OR "men 2b" OR cdhf12 OR "cdhf 12" OR cdhr16 OR "cdhr 16" OR "ret ele1" OR "retele 1" OR "rearranged during transfection protooncogene" OR mds OR prp10 OR "prp 10" OR hsh155 OR "hsh 155" OR prpf10 OR "prpf 10" OR sap155 OR "sap 155" OR sf3b155 OR "sf3b 155" OR "sf 3b 155" OR "splicing factor 3b subunit 1" OR sf3b1 OR "sf 3b 1" OR "sf3b 1") NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))):ti,ab,kw

#6 (((smad4 OR "smad 4" OR "deleted in pancreatic carcinoma 4" OR dpc4 OR "dpc 4" OR jip OR madh4 OR "madh 4" OR myhrs OR "smad family member 4" OR smarcb1 OR "smarcb 1" OR snf5 OR "snf 5" OR "integrase interactor 1" OR ini1 OR "ini 1" OR "malignant rhabdoid tumor suppressor" OR rdt OR css3 OR "css 3" OR snr1 OR "snr 1" OR baf47 OR "baf 47" OR mrd15 OR "mrd 15" OR rtps1 OR "rtps 1" OR sfh1p OR "sfh 1p" OR hsnfs OR snf5l1 OR "snf5l 1" OR "sn f5 l1" OR "sn f5l1" OR swnts1 OR "swnts 1" OR ppp1r144 OR "ppp1r 144" OR smo OR gx OR crjs OR smoh OR fzd11 OR "fzd 11" OR "homolog of smoothed" OR stk11 OR "stk 11" OR pjs OR lkb1 OR "lkb 1" OR hlkb1 OR "hlkb 1" OR tp53 OR "tp 53" OR bcc7 OR "bcc 7" OR lfs1 OR "lfs 1" OR bmfs5 OR "bmfs 5" OR p53 OR "p 53" OR "transformation related protein 53" OR trp53 OR "trp 53" OR "u2 af1" OR fp793 OR "fp 793" OR u2af35 OR "u2af 35" OR "u2 af 35" OR "u2 af35" OR u2afbp OR "u2 afbp" OR rnu2af1 OR "rnu2 af 1" OR u2af1 OR "u2af 1" OR "u2 af 1" OR u2 OR "u 2" OR "small nuclear rna auxiliary factor 1") NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))):ti,ab,kw

#7 (((he OR er OR esr OR era OR ago OR w OR bs OR met OR rn OR ld OR fat OR rn) NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((akt NEXT/4 3 NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR (("anaplastic lymphoma" NEXT/2 kinase NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5?-regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR (((arid OR "at rich interaction domain" OR "arid containing") NEXT/1 1a NEAR/3 (amplification OR chromosom* OR "copy

number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((brg1 NEAR/3 "250 kd*" NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((swi* NEXT/2 matrix* NEXT/2 actin NEXT/3 chromatin NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR (((("brca1 associated protein" OR bap) NEXT/1 "1" NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR (("discoidin domain receptor family" NEAR/2 "member 2" NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR mutant* OR mutat* OR allele OR allel* OR haplotyp* OR genotyp* OR mutation* OR indel OR indels OR frameshift* OR hypermutat* OR ((error* NEAR/3 (recombination OR replication))) OR (("gnaq g protein" NEAR/3 "alpha q" NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((harvey NEAR/2 sarcoma* NEAR/2 oncogene NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion)))):ti,ab,kw

#8 (((("isocitrate dehydrogenase" NEAR/2 nadp* NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR

nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((myeloid* NEXT/3 mixed NEXT/3 leukemia NEXT/2 ("2" OR "3" OR "4") NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((neuroblastoma NEXT/2 ras NEAR/4 oncogene NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((sf3b NEXT/1 "155 kd" NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((spliceosome associated protein" NEXT/1 155* NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR (("mothers against decapentaplegic" NEAR/3 "4" NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR (((sma OR mad) NEAR/3 related NEXT/3 "4" NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((swi* NEXT/4 matrix NEXT/3 actin NEXT/2 regulator NEXT/2 chromatin NEXT/2 "subfamily b" NEXT/1 "member 1" NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((smoothed NEXT/1 frizzled NEXT/2 receptor NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline*

OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion))) OR ((serine?threonine NEXT/2 kinase NEXT/2 11 NEAR/3 (amplification OR chromosom* OR "copy number" OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR "reading frame*" OR recombin* OR substitution OR frameshift* OR transition OR transversion)))):ti,ab,kw

#9 (mutant* OR mutat* OR allele OR allel* OR haplotyp* OR genotyp* OR mutation* OR indel OR indels OR frameshift* OR hypermutat*):ti,ab,kw

#10 (((error* NEAR/3 (recombination OR replication))) OR (((genomic OR gene OR genes OR genetic* OR oncogene* OR protooncogene* OR chromosom* OR exon* OR intron* OR codon* OR dna OR deoxyribonucleic OR germline* OR "5? regulatory" OR "3? untranslated" OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR dinucleotide* OR trinucleotide* OR intragenic OR "reading frame*") NEAR/3 (alter* OR chang* OR damag* OR disrupt* OR error* OR instab* OR defect* OR loss* OR missense OR nonsense OR frameshift* OR transition OR transversion OR substitution* OR deletion* OR insertion* OR duplication OR rearrangement* OR "copy number*" OR "repeat expansion")))) OR (((dna OR deoxyribonucleic) NEAR/3 sequenc*)):ti,ab,kw

#11 MeSH descriptor: [Mutation] explode all trees

#12 MeSH descriptor: [Genetic Variation] this term only

#13 MeSH descriptor: [Genetic Heterogeneity] explode all trees

#14 MeSH descriptor: [Polymorphism, Genetic] explode all trees

#15 MeSH descriptor: [Mutation Rate] explode all trees

#16 #1 OR #2 OR #3 OR #4 OR #5 OR #6 OR #7 OR #8 OR #9 OR #10 OR #11 OR #12 OR #13 OR #14 OR #15

#17 (((squamous OR epidermoid OR planocellular OR "prickle cell") NEAR/3 (cancer OR carcinoma* OR carcinogen*) NEAR/7 (skin* OR cutis* OR cutaneous OR epidermis OR dermis OR "stratum corneum" OR scalp* OR face OR faces OR facial OR auricle* OR cheek* OR eyelid* OR auricle* OR ear OR ears OR lip OR lips OR labia* OR palm* OR sole* OR aureol* OR keratinocyt*))) :ti,ab,kw

#18 (((scc OR csc) NEAR/7 (skin* OR cutis* OR cutaneous OR epidermis OR dermis OR "stratum corneum" OR scalp* OR face OR faces OR facial OR auricle* OR cheek* OR eyelid* OR auricle* OR ear OR ears OR lip OR lips OR labia* OR palm* OR sole* OR aureol* OR keratinocyt*))) :ti,ab,kw

#19 (squamous OR scc OR csc OR epidermoid OR planocellular OR "prickle cell"):ti

#20 MeSH descriptor: [Carcinoma, Squamous Cell] explode all trees

#21 #19 OR #20

#22 MeSH descriptor: [Skin Neoplasms] explode all trees

- #23 #21 AND #22
- #24 #17 OR #18 OR #23
- #25 #16 AND #24
- #26 MeSH descriptor: [Animals] explode all trees
- #27 MeSH descriptor: [Humans] explode all trees
- #28 #25 NOT (#26 NOT #27) 61

OID MEDLINE

1. ((akt3 or "akt 3" or "akt serine?threonine kinase 3" or mpph or mpph2 or "pkb gamma" or pkbγ or prkbγ or "rac pk gamma" or "stk 2" or "v akt" or "hgnc 393" or alk or cd246 or "cd 246" or nblst3 or "nblst 3" or apc or "adenomatous polyposis coli" or "deleted in polyposis 2.5" or "dp2.5" or "dp 2.5" or arid1a or "arid 1a" or eld or "b 120" or css2 or "css 2" or osa1 or "osa 1" or "p 270" or bm029 or "bm 029" or mrd14 or "mrd 14" or hosa1 or "hosa 1" or baf250 or "baf 250" or "baf 250a" or smarcf1 or "smarcf 1" or baf250a or "chromosome 1 open reading frame 4" or c1orf4 or "b120 gene" or b120 or p270 or bap1 or "bap 1" or uchl2 or "uchl 2" or hucep6 or "hucep 6" or hucep13 or "hucep 13" or cdh1 or "cdh 1" or cadherin or uvomorulin or "calcium-dependent adhesion protein" or "epithelial liver cell adhesion molecule" or uvo or cdhe or ecad or lcam or "arc 1" or arc1 or "bcds 1" or bcds1 or "cd 324" or cd324 or "cdk 4" or cdk4 or "cyclin dependent kinase 4" or "cmm 3" or cmm3 or "psk j3" or pskj3 or "pskj 3") adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5?-regulatory" or "3?-untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

2. (("cell division kinase 4" or cdkn2a or "cdkn 2a" or "cyclin-dependent kinase inhibitor 2a" or "cdk4 inhibitor" or "multiple tumor suppressor 1" or arf or mlm or p14 or "p 14" or p16 or "p 16" or p19 or "p 19" or cmm2 or "cmm 2" or ink4 or "ink 4" or mts1 or "mts 1" or tp16 or "tp 16" or cdk4i or "cdk 4i" or cdkn2 or "cdkn 2" or ink4a or "ink 4a" or p14arf or "p14 arf" or p19arf or "p19 arf" or p16ink4 or "p16 ink4" or p16ink4a or "p16 ink4a" or p16?ink4* or p16?ink4a or ddr2 or "discoidin domain receptor tyrosine kinase 2" or wrn or mig20a or "mig 20a" or tyro10 or "tyro 10" or "neurotrophic tyrosine kinase receptor-related 3" or ntrkr3 or "ntrkr 3" or "tyrosine kinase receptor related to neurotrophic trk" or tkt or egfr or "epidermal growth factor receptor" or "verb-b" or "verbb" or erb-b or erbb or erbb1 or "erbb 1" or her1 or

"her 1" or "species antigen 7" or sa7 or "sa 7" or mena or pig61 or "pig 61" or nisbd2 or "nisbd 2" or "erbB 2" or neu or her2* or "her 2*" or tkr1 or "tkr 1" or cd340 or "cd 340" or "mln 19" or mln19) adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5?-regulatory" or "3?-untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

3. (("erb b2" or erbb2 or "avian erythroblastic leukemia viral oncogene" or "neuroblastoma or glioblastoma derived oncogene" or ngl or erbb3* or "erbB b3" or "avian erythroblastic leukemia viral oncogene homolog 3" or her3 or "her 3" or ferlk or lccs2 or "lccs 2" or erbb3 or "mda bf 1" or "mda bf1" or "mdabf 1" or mdabf1 or "p180 erbb3" or p180erbb3 or "p45 serbb3" or p45serbb3 or "p85 serbb3" or p85serbb3 or "erb b4" or "erbB 4" or her4 or "her 4" or als19 or "als 19" or p180erbb4 or "p180 erbb4" or erbb4 or esr1 or "esr 1" or esra or estrr or nr3a1 or "estrogen receptor 1" or "estrogen receptor alpha" or "fat tumor suppressor" or fat1 or "fat 1" or me5 or "me 5" or cdhf7 or "cd hf7" or "cdhf 7" or cdhr8 or "cd hr8" or "cdhr 8" or hfat1 or "hfat 1" or "f box" or "wd40 domain protein 7" or fbxw7 or "fbxw 7" or fbw7 or "fbw 7" or fbxw6 or "fbxw 6" or fbw6 or "fbw 6" or fbxo30 or "fbxo 30" or fbx30 or "fbx 30" or "homolog of archipelago" or cdc4 or "cdc 4" or "homolog of sel10" or "homolog of sel-10" or fgfr2 or "fgfr 2" or "fibroblast growth factor receptor 2" or "fgf receptor" or "receptor-like protein tyrosine kinase 14" or bek or jws or bbds or cek3 or "cek 3" or cfd1 or "cfd 1" or ect1 or "ect 1" or kgfr or tk14 or "tk 14" or tk25 or "tk 25" or "bfr 1" or bfr1 or cd332 or "cd 332" or "k sam" or ksam or gata3 or "gata 3" or "gata binding protein 3") adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5?-regulatory" or "3?-untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

4. ((hdr or hdrs or "guanine nucleotide-binding protein" or "q polypeptide" or gnaq or "g protein" or "gq class" or gaq or sws or cmc1 or "cmc 1" or "g alpha q" or scg6 or "scg 6" or sgvi or "sg vi" or "gnas 1" or pita3 or "pita 3" or c20orf45 or gnas or gnas1 or aho or gsa or gsp or poh or gpsa or nesp or hnf1a or "hnf 1a" or lfb1 or "lfb 1" or tcf1 or "tcf 1" or hnf4a or "hnf 4a" or mody3 or "mody 3" or iddm20 or "hnf1 homeobox a" or "transcription factor 1" or "hepatocyte nuclear factor 1 alpha" or "hepatocyte nuclear factor 1" or hnf1 or "hnf 1" or "albumin proximal factor" or hras or hras1 or "hras 1" or rash1 or "rash 1" or p21?ras* or p21 or ctlo or kras or hamsv or kras2 or "kras 2" or rask2 or "rask 2" or "ki ras" or "c-bas?has" or p21ras or "c h ras" or chras or "c bas?has" or "c k ras" or ckras or "h rasidx" or "c ki ras" or ckiras or "c ha ras1" or idh1 or "idh 1" or "isocitrate dehydrogenase 1" or "peroxisomal isocitrate dehydrogenase" or picd or idpc or icdc or idh or idp or idcd or "hel 216" or hel216 or "hel s 26" or "hels 26" or hels26 or "hel s26" or idh2 or "idh 2" or "isocitrate dehydrogenase 2" or idpm or idhm or "icd m" or d2hga2 or "d2 hga2" or "d2hga 2" or jak2 or "jak 2" or jtk10 or "jtk 10" or theyt3 or "theyt 3" or "janus kinase 2" or kdr or "kinase insert domain receptor" or "tyrosine kinase growth factor receptor" or flk1 or "flk 1" or "vascular endothelial growth factor receptor" or vegfr2 or cd309 or "cd 309" or vegfr or keap1 or "keap 1" or "kelch-like ech-associated protein 1" or inrf2 or "inrf 2" or klhl19 or "klhl 19" or kit or "mast cell growth factor receptor" or "stem cell factor receptor" or scfr or fdc or ssm) adj3 (amplification or

chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5?-regulatory" or "3?-untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

5. ((sco1 or "sco 1" or sco5 or "sco 5" or sow3 or "sow 3" or cd117 or "cd 117" or "c kit" or "tr kit" or gsfsc01 or "gsfsc0 1" or gsfsc05 or "gsfsc0 5" or gsfscow3 or "gsfscow 3" or "lysine-specific methyltransferase 2c" or halr or mll3 or "mll 3" or klefs2 or "klefs 2" or kmt2c or "kmt 2c" or "lysine methyltransferase 2c" or kiaa1506 or "kiaa1 506" or kms or mll2 or "mll 2" or mll4 or "mll 4" or aad10 or "aad 10" or kabuk1 or "kabuk 1" or tnrc21 or "tnrc 21" or cagl114 or "cagl 114" or kmt2d or "kmt 2d" or "lysine methyltransferase 2d" or "all1 related" or "all 1 related" or alr or "mitogen-activated protein kinase" or "mapk?erk kinase 1" or mek1 or "mek 1" or cfc3 or "cfc 3" or mkk1 or "mkk 1" or mapkk1 or "mapkk 1" or prkmk1 or "prkmk 1" or map2k1 or "mitogen-activated protein kinase kinase 1" or cfc4 or "cfc 4" or mkk2 or "mkk 2" or mapkk2 or "mapkk 2" or prkmk2 or "prkmk 2" or map2k2 or "mitogen-activated protein kinase kinase 2" or "mapk?erk kinase 2" or mek2 or "mek 2" or "mitogen-activated kinase kinase kinase 1" or map3k1 or "map?erk kinase kinase 1" or mekk1 or "mekk 1" or mapkkk1 or "mapkkk 1" or "mek kinase" or mekk or srxy6 or "srxy 6" or auts9 or "auts 9" or rccp2 or "rccp 2" or "c met" or dfnb97 or "dfnb 97" or "met proto-oncogene" or "met oncogene" or "hepatocyte growth factor receptor" or hgfr) adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5?-regulatory" or "3?-untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

6. ((notch1 or "notch 1" or "notch receptor 1" or "translocation associated notch homolog" or tan1 or "tan 1" or hn1 or "hn 1" or aos5 or "aos 5" or aovd1 or "aovd 1" or ns6 or "ns 6" or cmns or ncms or alps4 or "alps 4" or "n ras" or nras1 or "nras 1" or nras or "pdgfr alpha" or cd140a or "cd 140a" or pdgfra or "platelet derived growth factor receptor alpha" or pdgfr2 or "pdgfr 2" or "phosphatidylinositol 3 kinase" or pik3ca or "p110 alpha" or "pi3k alpha" or "pik3 alpha" or mcm or cws5 or "cws 5" or mcap or pi3k or clapo or clove or mcmtc or "phosphatidylinositol 4?5 biphosphate 3 kinase catalytic subunit alpha" or pten or bzs or dec or cws1 or "cws 1" or glm2 or "glm 2" or mham or tep1 or "tep 1" or mmac1 or "mmac 1" or pten1 or "pten 1" or 10q23del or ptenbeta or "pten phosphatase and tensin homolog" or "mutated in multiple advanced cancers 1" or "phosphatase and tensin homolog deleted on chromosome 10" or rb1 or "rb 1" or "rb transcriptional corepressor 1" or rb or prb or osrc or pp110 or "pp 110" or ppp1r130 or "ppp1r 130" or "p105 rb" or ret or ptc or mtc1 or "mtc 1" or hscr1 or "hscr 1" or men2a or "men 2a" or men2b or "men 2b" or cdhf12 or "cdhf 12" or cdhr16 or "cdhr 16" or "ret ele1" or "retele 1" or "rearranged during transfection protooncogene" or mds or prp10 or "prp 10" or hsh155 or "hsh 155" or prpf10 or "prpf 10" or sap155 or "sap 155" or sf3b155 or "sf3b 155" or "sf 3b 155" or "splicing factor 3b subunit 1" or sf3b1 or "sf 3b 1" or "sf3b 1" or smad4 or "smad 4" or "deleted in pancreatic carcinoma 4" or dpc4 or "dpc 4" or jip) adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or

intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

7. ((madh4 or "madh 4" or myhrs or "smad family member 4" or smarcb1 or "smarcb 1" or snf5 or "snf 5" or "integrase interactor 1" or ini1 or "ini 1" or "malignant rhabdoid tumor suppressor" or rdt or css3 or "css 3" or snr1 or "snr 1" or baf47 or "baf 47" or mrd15 or "mrd 15" or rtps1 or "rtps 1" or sfh1p or "sfh 1p" or hsnfs or snf511 or "snf51 1" or "sn f5 11" or "sn f511" or swnts1 or "swnts 1" or ppp1r144 or "ppp1r 144" or smo or gx or crjs or smoh or fzd11 or "fzd 11" or "homolog of smoothed" or stk11 or "stk 11" or pjs or lkb1 or "lkb 1" or hlkb1 or "hlkb 1" or tp53 or "tp 53" or bcc7 or "bcc 7" or lfs1 or "lfs 1" or bmfs5 or "bmfs 5" or p53 or "p 53" or "transformation related protein 53" or trp53 or "trp 53" or "u2 af1" or fp793 or "fp 793" or u2af35 or "u2af 35" or "u2 af 35" or "u2 af35" or u2afbp or "u2 afbp" or rnu2af1 or "rnu2 af 1" or u2af1 or "u2af 1" or "u2 af 1" or u2 or "u 2" or "small nuclear rna auxiliary factor 1") adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

8. (mutant* or mutat* or allele or allel* or haplotyp* or genotyp* or mutation* or indel or indels or frameshift* or hypermutat*).ab,ti.

9. (error* adj3 (recombination or replication)).ab,ti.

10. ((genomic or gene or genes or genetic* or oncogene* or protooncogene* or chromosom* or exon* or intron* or codon* or dna or deoxyribonucleic or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or dinucleotide* or trinucleotide* or intragenic or "reading frame*") adj3 (alter* or chang* or damag* or disrupt* or error* or instab* or defect* or loss* or missense or nonsense or frameshift* or transition or transversion or substitution* or deletion* or insertion* or duplication or rearrangement* or "copy number*" or "repeat expansion")).ab,ti.

11. ((dna or deoxyribonucleic) adj3 sequenc*).ab,ti.

12. ((hE or ER or ESR or Era or ago or W or Bs or met or RN or LD or FAT or RN) adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

13. (AKT adj4 (("erb b2" or erbb2 or "avian erythroblastic leukemia viral oncogene" or "neuroblastoma or glioblastoma derived oncogene" or ngl or erbb3* or "erbb b3" or "avian erythroblastic leukemia viral oncogene homolog 3" or her3 or "her 3" or ferlk or lccs2 or "lccs 2" or erbb3 or "mda bf 1" or "mda bf1" or "mdabf 1" or mdabf1 or "p180 erbb3" or p180erbb3 or "p45 serbb3" or p45serbb3 or "p85 serbb3" or p85serbb3 or "erb b4" or "erbb 4" or her4 or "her 4" or als19 or "als 19" or p180erbb4 or "p180 erbb4" or erbb4 or esr1 or "esr 1" or esra or estr or nr3a1 or "estrogen receptor 1" or "estrogen receptor alpha" or "fat tumor suppressor" or fat1 or "fat 1" or me5 or "me 5" or cdhf7 or "cd hf7" or "cdhf 7" or cdhr8 or "cd hr8" or "cdhr 8" or hfat1 or "hfat 1" or "f box" or "wd40 domain protein 7" or fbxw7 or "fbxw 7" or fbw7 or

"fbw 7" or fbw6 or "fbw 6" or fbw6 or "fbw 6" or fbw30 or "fbw 30" or fbw30 or "fbw 30" or "homolog of archipelago" or cdc4 or "cdc 4" or "homolog of sel10" or "homolog of sel-10" or fgfr2 or "fgfr 2" or "fibroblast growth factor receptor 2" or "fgf receptor" or "receptor-like protein tyrosine kinase 14" or bek or jws or bbds or cek3 or "cek 3" or cfd1 or "cfd 1" or ect1 or "ect 1" or kgfr or tk14 or "tk 14" or tk25 or "tk 25" or "bfr 1" or bfr1 or cd332 or "cd 332" or "k sam" or ksam or gata3 or "gata 3" or "gata binding protein 3") adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5?-regulatory" or "3?-untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)) adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

14. ("anaplastic lymphoma" adj2 kinase adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

15. ((arid or AT-rich-interaction-domain or arid-containing) adj 1a adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

16. (brg1 adj3 "250 kd*" adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

17. (SWI* adj2 matrix* adj2 actin adj3 chromatin adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

18. (("BRCA1 ASSOCIATED PROTEIN" or BAP) adj "1" adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or

deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

19. ("GNAQ G protein" adj3 "alpha q" adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

20. (harvey adj2 sarcoma* adj2 oncogene adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

21. ("ISOCITRATE DEHYDROGENASE" adj2 NADP* adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

22. (MYELOID* adj3 MIXED adj3 LEUKEMIA adj2 ("2" or "3" or "4") adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

23. (NEUROBLASTOMA adj2 RAS adj4 ONCOGENE adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

24. (SF3B adj 155-KD adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

25. ("SPLICEOSOME ASSOCIATED PROTEIN" adj 155* adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

26. ("MOTHERS AGAINST DECAPENTAPLEGIC" adj3 "4" adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

27. ((SMA or MAD) adj3 related adj3 "4" adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

28. (SWI* adj4 MATRIX adj3 ACTIN adj2 REGULATOR adj2 CHROMATIN adj2 "SUBFAMILY B" adj "MEMBER 1" adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3 -untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

29. (smoothened adj frizzled adj2 receptor adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

30. (SERINE?THREONINE adj2 KINASE adj2 ((dna or deoxyribonucleic) adj3 sequenc*) adj3 (amplification or chromosom* or "copy number" or deletion or duplicat* or exon* or insertion* or intron* or dna or deoxyribonucleic or gene or genes or oncogene* or protooncogene* or genetic* or genom* or genotyp* or germline* or "5? regulatory" or "3? untranslated" or promoter* or splicing or deoxynucleotide* or nucleotide* or intragenic or missense or nonsense or "reading frame*" or recombin* or substitution or frameshift* or transition or transversion)).ab,ti.

31. exp Mutation/

32. genetic variation/ or exp genetic heterogeneity/ or exp polymorphism, genetic/

33. exp mutation rate/

34. 1 or 2 or 3 or 4 or 5 or 6 or 7 or 8 or 9 or 10 or 11 or 12 or 13 or 14 or 15 or 16 or 17 or 18 or 19 or 20 or 21 or 22 or 23 or 24 or 25 or 26 or 27 or 28 or 29 or 30 or 31 or 32 or 33

35. ((scc or csc) adj7 (skin* or cutis* or cutaneous or epidermis or dermis or "stratum corneum" or scalp* or face or faces or facial or auricle or cheek or eyelid or auricle* or ear or ears or lip or lips or labia* or palm* or sole* or aureol* or keratinocyt*)).ab,ti.

36. ((squamous or epidermoid or planocellular or "prickle cell") adj3 (cancer or carcinoma* or carcinogen*) adj7 (skin* or cutis* or cutaneous or epidermis or dermis or "stratum corneum" or scalp* or face or faces or facial or auricle or cheek or eyelid or auricle* or ear or ears or lip or lips or labia* or palm* or sole* or aureol* or keratinocyt*)).ab,ti.

37. exp Carcinoma, Squamous Cell/
38. (squamous or scc or csc or epidermoid or planocellular or "prickle cell").ti.
39. 37 or 38
40. exp Skin Neoplasms/
41. 39 and 40
42. 35 or 36 or 41
43. 34 and 42
44. limit 43 to english language
45. exp Animals/
46. exp Humans/
47. 44 not (45 not 46)

EMBASE

No. Query Results

#5 (#1 OR #2 OR #3) AND #4 1446

#4 (('squamous cell carcinoma'/exp OR squamous:ti OR scc:ti OR csc:ti OR epidermoid:ti OR planocellular:ti OR 'prickle cell':ti) AND 'skin cancer'/exp/mj OR (((squamous OR epidermoid OR planocellular OR 'prickle cell') NEAR/3 (cancer OR carcinoma* OR carcinogen*) NEAR/7 (skin* OR cutis* OR cutaneous OR epidermis OR dermis OR 'stratum corneum' OR scalp* OR face OR faces OR facial OR auricle* OR cheek* OR eyelid* OR auricle* OR ear OR ears OR lip OR lips OR labia* OR palm* OR sole* OR aureol* OR keratinocyt*)):ab,kw,ti) OR (((scc OR csc) NEAR/7 (skin* OR cutis* OR cutaneous OR epidermis OR dermis OR 'stratum corneum' OR scalp* OR face OR faces OR facial OR auricle* OR cheek* OR eyelid* OR auricle* OR ear OR ears OR lip OR lips OR labia* OR palm* OR sole* OR aureol* OR keratinocyt*)):ab,kw,ti)) NOT ([animals]/lim NOT [humans]/lim) AND [english]/lim NOT 'conference abstract'/it 13277

#3 (('isocitrate dehydrogenase' NEAR/2 nadp* NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((myeloid* NEXT/3 mixed NEXT/3 leukemia NEXT/2 ('2' OR '3' OR '4') NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((neuroblastoma NEXT/2 ras NEAR/4 oncogene NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR

deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((sf3b NEXT/1 '155 kd' NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR (('spliceosome-associated protein' NEXT/1 155* NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR (('mothers against decapentaplegic' NEAR/3 '4' NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR (((sma OR mad) NEAR/3 related NEXT/3 '4' NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((swi* NEXT/4 matrix NEXT/3 actin NEXT/2 regulator NEXT/2 chromatin NEXT/2 'subfamily b' NEXT/1 'member 1' NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((smoothed NEXT/1 frizzled NEXT/2 receptor NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((serine?threonine NEXT/2 kinase NEXT/2 11 NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti)

#2 (((he OR er OR esr OR era OR ago OR w OR bs OR met OR rn OR ld OR fat OR rn) NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((akt NEXT/4 3 NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR (('anaplastic lymphoma' NEXT/2 kinase NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR (((arid OR 'at rich interaction domain' OR 'arid containing') NEXT/1 1a NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((brg1 NEAR/3 '250 kd*' NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((swi* NEXT/2 matrix* NEXT/2 actin NEXT/3 chromatin NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR (((brca1-associated protein' OR bap) NEXT/1 '1' NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR (('discoidin domain receptor family' NEAR/2 'member 2' NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR

missense OR nonsense OR 'reading frame*' OR recomb* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR (('gnaq g protein' NEAR/3 'alpha q' NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recomb* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR ((harvey NEAR/2 sarcoma* NEAR/2 oncogene NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recomb* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) 31289

#1 (((akt3 OR 'akt 3' OR 'akt serine/threonine kinase 3' OR mpph OR mpph2 OR 'pkb gamma' OR 'pkb γ' OR pkby OR pkbγ OR prkbg OR 'rac pk gamma' OR 'rac pk γ' OR 'rac pky' OR 'racpkγ' OR 'racpk γ' OR 'rac γ' OR 'racγ' OR 'stk 2' OR 'v akt' OR 'hgnc 393' OR alk OR cd246 OR 'cd 246' OR nblst3 OR 'nblst 3' OR apc OR 'adenomatous polyposis coli' OR 'deleted in polyposis 2.5' OR dp2.5 OR 'dp 2.5' OR arid1a OR 'arid 1a' OR eld OR 'b 120' OR css2 OR 'css 2' OR osa1 OR 'osa 1' OR 'p 270' OR bm029 OR 'bm 029' OR mrd14 OR 'mrd 14' OR hosa1 OR 'hosa 1' OR baf250 OR 'baf 250' OR 'baf 250a' OR smarcf1 OR 'smarcf 1' OR baf250a OR 'chromosome 1 open reading frame 4' OR c1orf4 OR 'b120 gene' OR b120 OR p270 OR bap1 OR 'bap 1' OR uchl2 OR 'uchl 2' OR hucep6 OR 'hucep 6' OR hucep13 OR 'hucep 13' OR cdh1 OR 'cdh 1' OR cadherin OR uvomorulin OR 'calcium-dependent adhesion protein' OR 'epithelial liver cell adhesion molecule' OR uvo OR cdhe OR ecad OR lcam OR 'arc 1' OR arc1 OR 'bcds 1' OR bcds1 OR 'cd 324' OR cd324 OR 'cdk 4' OR cdk4 OR 'cyclin dependent kinase 4' OR 'cmm 3' OR cmm3 OR 'psk j3' OR pskj3 OR 'pskj 3' OR 'cell division kinase 4' OR cdkn2a OR 'cdkn 2a' OR 'cyclin-dependent kinase inhibitor 2a' OR 'cdk4 inhibitor' OR 'multiple tumor suppressor 1' OR arf OR mlm OR p14 OR 'p 14' OR p16 OR 'p 16' OR p19 OR 'p 19' OR cmm2 OR 'cmm 2' OR ink4 OR 'ink 4' OR mts1 OR 'mts 1' OR tp16 OR 'tp 16' OR cdk4i OR 'cdk 4i' OR cdkn2 OR 'cdkn 2' OR ink4a OR 'ink 4a' OR p14arf OR 'p14 arf' OR p19arf OR 'p19 arf' OR p16ink4 OR 'p16 ink4' OR p16ink4a OR 'p16 ink4a' OR p16\$ink4* OR p16\$ink4a OR ddr2 OR 'discoidin domain receptor tyrosine kinase 2' OR wrcn OR mig20a OR 'mig 20a' OR tyro10 OR 'tyro 10' OR 'neurotrophic tyrosine kinase receptor-related 3' OR ntrkr3 OR 'ntrkr 3' OR 'tyrosine kinase receptor related to neurotrophic trk' OR tkt OR egfr OR 'epidermal growth factor receptor' OR 'verb-b' OR 'verbb' OR 'erb b' OR erbb OR erbb1 OR 'erbb 1' OR her1 OR 'her 1' OR 'species antigen 7' OR sa7 OR 'sa 7' OR mena OR pig61 OR 'pig 61' OR nisbd2 OR 'nisbd 2' OR 'erbb 2' OR neu OR her2* OR 'her 2*' OR tkr1 OR 'tkr 1' OR cd340 OR 'cd 340' OR 'mln 19' OR mln19 OR 'erb b2' OR erbb2 OR 'avian erythroblastic leukemia viral oncogene' OR 'neuroblastoma or glioblastoma derived oncogene' OR ngl OR erbb3* OR 'erbb b3' OR 'avian erythroblastic leukemia viral oncogene homolog 3' OR her3 OR 'her 3' OR ferlk OR lccs2 OR 'lccs 2' OR erbb3 OR 'mda bf 1' OR 'mda bf1' OR 'mdabf 1' OR mdabf1 OR 'p180 erbb3' OR p180erbb3 OR 'p45 serbb3' OR p45serbb3 OR 'p85 serbb3' OR p85serbb3 OR 'erb b4' OR 'erbb 4' OR her4 OR 'her 4' OR als19 OR 'als 19' OR p180erbb4 OR 'p180 erbb4' OR erbb4 OR esr1 OR 'esr 1' OR esra OR estrr OR nr3a1 OR 'estrogen receptor 1' OR 'estrogen receptor alpha' OR 'estrogen receptor α' OR 'fat tumor suppressor' OR fat1 OR 'fat 1' OR me5 OR 'me 5' OR cdhf7 OR 'cd hf7' OR 'cdhf 7' OR cdhr8 OR 'cd hr8' OR 'cdhr 8' OR hfat1 OR 'hfat 1' OR 'f box' OR 'wd40 domain protein 7' OR fbxw7 OR 'fbxw 7' OR fbw7 OR 'fbw 7' OR fbxw6 OR 'fbxw 6' OR fbw6 OR 'fbw 6' OR fbxo30 OR 'fbxo 30' OR fbx30 OR 'fbx 30'

OR 'homolog of archipelago' OR cdc4 OR 'cdc 4' OR 'homolog of sel10' OR 'homolog of sel-10' OR fgfr2 OR 'fgfr 2' OR 'fibroblast growth factor receptor 2' OR 'fgf receptor' OR 'receptor-like protein tyrosine kinase 14' OR bek OR jws OR bbds OR cek3 OR 'cek 3' OR cfd1 OR 'cfd 1' OR ect1 OR 'ect 1' OR kgfr OR tk14 OR 'tk 14' OR tk25 OR 'tk 25' OR bfr1 OR 'bfr 1' OR cd332 OR 'cd 332' OR 'k sam' OR ksam OR gata3 OR 'gata 3' OR 'gata binding protein 3' OR hdr OR hds OR 'guanine nucleotide-binding protein' OR 'g polypeptide' OR gnaq OR 'g protein' OR 'gq class' OR 'g α q' OR g α q OR gaq OR sws OR cmc1 OR 'cmc 1' OR 'g alpha q' OR 'g α q' OR scg6 OR 'scg 6' OR sgvi OR 'sg vi' OR gnas1 OR pita3 OR 'pita 3' OR c20orf45 OR gnas OR gnas1 OR aho OR gsa OR gsp OR poh OR gpsa OR nesp OR hnf1a OR 'hnf 1a' OR lfb1 OR 'lfb 1' OR tcf1 OR 'tcf 1' OR hnf4a OR 'hnf 4a' OR mody3 OR 'mody 3' OR iddm20 OR 'hnf1 homeobox a' OR 'transcription factor 1' OR 'hepatocyte nuclear factor 1 alpha' OR 'hepatocyte nuclear factor 1 α ' OR 'hepatocyte nuclear factor 1 α ' OR 'hepatocyte nuclear factor 1' OR hnf1 OR 'hnf 1' OR 'albumin proximal factor' OR hras OR hras1 OR 'hras 1' OR rash1 OR 'rash 1' OR p21^{ras} OR p21 OR ctlo OR kras OR hamsv OR kras2 OR 'kras 2' OR rask2 OR 'rask 2' OR 'ki ras' OR 'c-bas/has' OR p21ras OR 'c h ras' OR chras OR 'c bas/has' OR 'c k ras' OR ckras OR 'h rasid' OR 'c ki ras' OR ckiras OR 'c ha ras1' OR idh1 OR 'idh 1' OR 'isocitrate dehydrogenase 1' OR 'peroxisomal isocitrate dehydrogenase' OR picd OR idpc OR icdc OR idh OR idp OR idcd OR 'hel 216' OR hel216 OR 'hel s 26' OR 'hels 26' OR hels26 OR 'hel s26' OR idh2 OR 'idh 2' OR 'isocitrate dehydrogenase 2' OR idpm OR idhm OR 'icd m' OR d2hga2 OR 'd2 hga2' OR 'd2hga 2' OR jak2 OR 'jak 2' OR jtk10 OR 'jtk 10' OR thcyt3 OR 'thcyt 3' OR janus kinase 2' OR kdr OR 'kinase insert domain receptor' OR 'tyrosine kinase growth factor receptor' OR flk1 OR 'flk 1' OR 'vascular endothelial growth factor receptor' OR vegfr2 OR cd309 OR 'cd 309' OR vegfr OR keap1 OR 'keap 1' OR 'kelch-like ech-associated protein 1' OR inrf2 OR 'inrf 2' OR klhl19 OR 'klhl 19' OR kit OR 'mast cell growth factor receptor' OR 'stem cell factor receptor' OR scfr OR fdc OR ssm OR sco1 OR 'sco 1' OR sco5 OR 'sco 5' OR sow3 OR 'sow 3' OR cd117 OR 'cd 117' OR 'c kit' OR 'tr kit' OR gsfsc1 OR 'gsfsco 1' OR gsfsc5 OR 'gsfsco 5' OR gsfscow3 OR 'gsfscow 3' OR 'lysine-specific methyltransferase 2c' OR halr OR mll3 OR 'mll 3' OR klefs2 OR 'klefs 2' OR kmt2c OR 'kmt 2c' OR 'lysine methyltransferase 2c' OR kiaa1506 OR 'kiaa 1506' OR kms OR mll2 OR 'mll 2' OR mll4 OR 'mll 4' OR aad10 OR 'aad 10' OR kabuk1 OR 'kabuk 1' OR tnrc21 OR 'tnrc 21' OR cagl114 OR 'cagl 114' OR kmt2d OR 'kmt 2d' OR 'lysine methyltransferase 2d' OR 'all1 related' OR 'all 1 related' OR alr OR 'mitogen-activated protein kinase' OR 'mapk ζ erk kinase 1' OR mek1 OR 'mek 1' OR cfc3 OR 'cfc 3' OR mkk1 OR 'mkk 1' OR mapkk1 OR 'mapkk 1' OR prkmk1 OR 'prkmk 1' OR map2k1 OR 'mitogen-activated protein kinase kinase 1' OR cfc4 OR 'cfc 4' OR mkk2 OR 'mkk 2' OR mapkk2 OR 'mapkk 2' OR prkmk2 OR 'prkmk 2' OR map2k2 OR 'mitogen-activated protein kinase kinase 2' OR 'mapk ζ erk kinase 2' OR mek2 OR 'mek 2' OR 'mitogen-activated kinase kinase kinase 1' OR map3k1 OR 'map ζ erk kinase kinase 1' OR mekk1 OR 'mekk 1' OR mapkkk1 OR 'mapkkk 1' OR 'mek kinase' OR mekk OR srxy6 OR 'srxy 6' OR auts9 OR 'auts 9' OR rccp2 OR 'rccp 2' OR 'c met' OR dfnb97 OR 'dfnb 97' OR 'met proto-oncogene' OR 'met oncogene' OR 'hepatocyte growth factor receptor' OR hgfr OR notch1 OR 'notch 1' OR 'notch receptor 1' OR 'translocation-associated notch homolog' OR tan1 OR 'tan 1' OR hn1 OR 'hn 1' OR aos5 OR 'aos 5' OR aovd1 OR 'aovd 1' OR ns6 OR 'ns 6' OR cmns OR ncms OR alps4 OR 'alps 4' OR 'n ras' OR nras1 OR 'nras 1' OR nras OR pdgfra OR 'pdgfr α ' OR pdgfralpha OR 'pdgfr alpha' OR cd140a OR 'cd 140a' OR pdgfra OR 'platelet derived growth factor receptor alpha' OR 'platelet derived growth factor receptor α ' OR pdgfr2 OR 'pdgfr 2' OR 'phosphatidylinositol 3-kinase' OR pik3ca OR 'p110 alpha' OR 'p110 α ' OR p110 α OR 'pi3k alpha' OR 'pi3k α ' OR pi3k α OR 'pik3 alpha' OR 'pik3 α ' OR pik3 α OR mcm OR cws5 OR 'cws 5' OR mcap OR pi3k OR clapo OR clove OR mcmtc OR 'phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit alpha' OR 'phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit α ' OR pten OR bzs OR dec OR cws1 OR 'cws 1' OR glm2 OR 'glm 2' OR mham OR tep1

OR 'tep 1' OR mmac1 OR 'mmac 1' OR pten1 OR 'pten 1' OR 10q23del OR ptenbeta OR ptenβ OR 'pten phosphatase and tensin homolog' OR 'mutated in multiple advanced cancers 1' OR 'phosphatase and tensin homolog deleted on chromosome 10' OR rb1 OR 'rb 1' OR 'rb transcriptional corepressor 1' OR rb OR prb OR osrc OR pp110 OR 'pp 110' OR ppp1r130 OR 'ppp1r 130' OR 'p105 rb' OR ret OR ptc OR mtc1 OR 'mtc 1' OR hscr1 OR 'hscr 1' OR men2a OR 'men 2a' OR men2b OR 'men 2b' OR cdhf12 OR 'cdhf 12' OR cdhr16 OR 'cdhr 16' OR 'ret ele1' OR 'retele 1' OR 'rearranged during transfection protooncogene' OR mds OR prp10 OR 'prp 10' OR hsh155 OR 'hsh 155' OR prpf10 OR 'prpf 10' OR sap155 OR 'sap 155' OR sf3b155 OR 'sf3b 155' OR 'sf 3b 155' OR 'splicing factor 3b subunit 1' OR sf3b1 OR 'sf 3b 1' OR 'sf3b 1' OR smad4 OR 'smad 4' OR 'deleted in pancreatic carcinoma 4' OR dpc4 OR 'dpc 4' OR jip OR madh4 OR 'madh 4' OR myhrs OR 'smad family member 4' OR smarcb1 OR 'smarcb 1' OR snf5 OR 'snf 5' OR 'integrase interactor 1' OR ini1 OR 'ini 1' OR 'malignant rhabdoid tumor suppressor' OR rdt OR css3 OR 'css 3' OR snr1 OR 'snr 1' OR baf47 OR 'baf 47' OR mrd15 OR 'mrd 15' OR rtps1 OR 'rtps 1' OR sfh1p OR 'sfh 1p' OR hsnfs OR snf5l1 OR 'snf5l 1' OR 'sn f5 l1' OR 'sn f5l1' OR swnts1 OR 'swnts 1' OR ppp1r144 OR 'ppp1r 144' OR smo OR gx OR crjs OR smoh OR fzd11 OR 'fzd 11' OR 'homolog of smoothed' OR stk11 OR 'stk 11' OR pjs OR lkb1 OR 'lkb 1' OR hlkb1 OR 'hlkb 1' OR tp53 OR 'tp 53' OR bcc7 OR 'bcc 7' OR lfs1 OR 'lfs 1' OR bmfs5 OR 'bmfs 5' OR p53 OR 'p 53' OR 'transformation-related protein 53' OR trp53 OR 'trp 53' OR 'u2 af1' OR fp793 OR 'fp 793' OR u2af35 OR 'u2af 35' OR 'u2 af 35' OR 'u2 af35' OR u2afbp OR 'u2 afbp' OR rnu2af1 OR 'rnu2 af 1' OR u2af1 OR 'u2af 1' OR 'u2 af 1' OR u2 OR 'u 2' OR 'small nuclear rna auxiliary factor 1') NEAR/3 (amplification OR chromosom* OR 'copy number' OR deletion OR duplicat* OR exon* OR insertion* OR intron* OR dna OR deoxyribonucleic OR gene OR genes OR oncogene* OR protooncogene* OR genetic* OR genom* OR genotyp* OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR intragenic OR missense OR nonsense OR 'reading frame*' OR recombin* OR substitution OR frameshift* OR transition OR transversion)):ab,kw,ti) OR mutant*:ab,kw,ti OR mutat*:ab,kw,ti OR allele:ab,kw,ti OR allel*:ab,kw,ti OR haplotyp*:ab,kw,ti OR genotyp*:ab,kw,ti OR mutation*:ab,kw,ti OR indel:ab,kw,ti OR indels:ab,kw,ti OR frameshift*:ab,kw,ti OR hypermutat*:ab,kw,ti OR ((error* NEAR/3 (recombination OR replication)):ab,kw,ti) OR (((genomic OR gene OR genes OR genetic* OR oncogene* OR protooncogene* OR chromosom* OR exon* OR intron* OR codon* OR dna OR deoxyribonucleic OR germline* OR '5\$-regulatory' OR '3\$-untranslated' OR promoter* OR splicing OR deoxynucleotide* OR nucleotide* OR dinucleotide* OR trinucleotide* OR intragenic OR 'reading frame*') NEAR/3 (alter* OR chang* OR damag* OR disrupt* OR error* OR instab* OR defect* OR loss* OR missense OR nonsense OR frameshift* OR transition OR transversion OR substitution* OR deletion* OR insertion* OR duplication OR rearrangement* OR 'copy number*' OR 'repeat expansion')):ab,kw,ti) OR (((dna OR deoxyribonucleic) NEAR/3 sequenc*):ab,kw,ti) OR 'gene mutation'/exp OR 'genomic instability'/exp OR 'germline mutation'/exp OR 'induced mutation'/exp OR 'mutagenesis'/exp OR 'mutant'/de OR 'deletion mutant'/exp OR 'lethal mutant'/exp OR 'temperature sensitive mutant'/exp OR 'revertant'/exp OR 'mutation rate'/exp OR 'somatic hypermutation'/exp OR 'spontaneous mutation'/exp 2290000