

Supplementary Table 1. Key terms used to prioritize genetic variants with functional annotations and/or bibliography related to cancer. Functional annotations listed corresponded to those selected from Gene Ontology, KEGG and REACTOME, whereas the Bibliography category referred to terms from NCBI gene summary and Gene Reference Into Function (GeneRIF). Noteworthy, other annotations containing listed key terms were also selected (e.g. “breast cancer” since it contains “cancer”).

Gene ontology biological process terms	
Apoptosis and cell death	Abortive mitotic cell cycle
<u>Apoptotic process</u> Execution phase of apoptosis Apoptotic signaling pathway Mitochondrial outer membrane permeabilization Release of cytochrome c from mitochondria Apoptosome assembly <u>Cell death</u> Programmed cell death Autophagic cell death Macroautophagy Microautophagy Regulation of autophagy Cytolysis Necrotic cell death	Mitotic spindle organization Mitotic nuclear envelope disassembly Mitotic nuclear envelope reassembly Establishment of mitotic spindle localization Mitosis Mitotic prophase Mitotic metaphase Mitotic anaphase Mitotic telophase Mitotic prometaphase Mitotic sister chromatid segregation Spindle assembly involved in mitosis Mitotic spindle assembly checkpoint Mitotic centrosome separation Cell cycle arrest Cell cycle checkpoint DNA integrity checkpoint Spindle assembly checkpoint
Cell adhesion	Cell differentiation
Cell-cell adhesion Homophilic cell adhesion Cell-cell junction assembly Cell-substrate junction assembly Regulation of cell junction assembly Adherens junction assembly Cell adhesion mediated by integrin Cell-substrate adhesion Regulation of cell adhesion Tight junction assembly	Cell differentiation Epithelial cell differentiation G1 to G0 transition involved in cell differentiation Mesodermal cell differentiation Stem cell differentiation Cell development Cell fate commitment Negative regulation of cell differentiation Positive regulation of cell differentiation Regulation of epithelial cell differentiation
Cell cycle	Cell growth
Cell cycle Cell cycle phase Cell cycle phase transition Mitotic cell cycle	Cell growth

Developmental cell growth
Regulation of cell proliferation
Regulation of epithelial cell proliferation
Cellular response to DNA damage stimulus
DNA repair
Single strand break repair
Mismatch repair
Postreplication repair
Double-strand break repair
DNA replication proofreading
Non-recombinational repair
Base-excision repair
Nucleotide-excision repair
UV-damage excision repair
Interstrand cross-link repair
DNA damage checkpoint
DNA damage response, detection of DNA damage
DNA damage induced protein phosphorylation
Replication fork processing
Telomere maintenance in response to DNA damage
Intrinsic apoptotic signaling pathway in response to DNA damage
Signal transduction in response to DNA damage
Chromatin
Chromatin organization
Helicase activity
Maintenance of DNA methylation
Chromosome organization
Kinetochore organization
Chromosome condensation
Chromosome decondensation
Chromosome breakage
Telomere organization
Maintenance of DNA repeat elements
Sister chromatid cohesion
Regulation of cohesin localization to chromatin
Centromere complex assembly
Regulation of chromosome organization
Maintenance of chromatin silencing
Telomere maintenance
Maintenance of fidelity involved in DNA-dependent DNA replication
DNA protection
Chromatin remodeling
Pathways
Wnt receptor signaling pathway
BMP signaling pathway
SMAD protein signal transduction

SMAD protein complex assembly
Negative regulation of MAPKKK cascade
Negative regulation of MAP kinase activity
Regulation of JUN kinase activity
Negative regulation of JNK cascade
Insulin receptor signaling pathway
Cellular response to TGF-beta stimulus
Cellular response to TGF-beta stimulus
Transforming growth factor beta receptor signaling pathway
Response to transforming growth factor beta stimulus
Epidermal growth factor receptor signaling pathway
Notch signaling pathway
Positive regulation of transcription of Notch receptor target
Notch receptor processing, ligand-dependent
Negative regulation of protein kinase B signaling cascade
Anterior/posterior pattern formation
Netrin-activated signaling pathway
Regulation of TOR signaling cascade
Target of rapamycin signaling pathway
Regulation of phosphatidylinositol 3-kinase cascade
NF-kappaB import into nucleus
Regulation of NIK/NF-kappaB cascade
Regulation of p52-dependent NF-kappaB signaling
KEGG pathway terms
Base excision repair
Nucleotide excision repair
Mismatch repair
Homologous recombination
Non-homologous end-joining
Cell cycle checkpoint
Metabolism of xenobiotics by cytochrome P450
Benzoate degradation
Aminobenzoate degradation
Fluorobenzoate degradation
Chloroalkane and chloroalkene degradation
Chlorocyclohexane and chlorobenzene degradation
Toluene degradation
Xylene degradation
Nitrotoluene degradation
Ethylbenzene degradation
Styrene degradation
Atrazine degradation
Caprolactam degradation
DDT degradation
Bisphenol degradation
Dioxin degradation

Naphthalene degradation
 Polycyclic aromatic hydrocarbon degradation
 Furfural degradation
 Steroid degradation
 Metabolism of xenobiotics by cytochrome P450
 Focal adhesion
 Adherens junction
 Tight junction
 Gap junction
 Purine metabolism
 Pyrimidine metabolism
 Wnt signaling pathway
 Notch signaling pathway
 TGF-beta signaling pathway
 Insulin signaling pathway
 PI3K-Akt signaling pathway
 Mucin type O-Glycan biosynthesis

Integrin cell surface interactions
 Adherens junctions interactions
 Cell-Cell communication
 Telomere maintenance
 Extension of Telomeres
 Chromosome maintenance
 Signaling by BMP
 Signaling by TGF beta
 Signaling by Wnt
 Beta-catenin phosphorylation cascade
 Degradation of beta-catenin by the destruction complex
 PKB-mediated events
 TGF-beta receptor signaling
 Constitutive PI3K/AKT Signaling in Cancer
 Chromosome maintenance
 O-linked glycosylation of mucins

Reactome terms

DNA repair
 Base excision repair
 Cleavage of the damaged purine
 Depurination
 Removal of DNA patch containing abasic residue
 Resolution of AP sites via the single-nucleotide replacement pathway
 Resolution of Abasic Sites (AP sites), organism-specific biosystem
 Gap-filling DNA repair synthesis and ligation in GG-NER
 Gap-filling DNA repair synthesis and ligation in TC-NER
 Transcription-coupled NER (TC-NER)
 Repair synthesis for gap-filling by DNA polymerase in TC-NER
 Apoptosis
 Apoptotic execution phase
 Apoptotic cleavage of cellular proteins
 Mitotic anaphase
 M phase
 G1/S transition
 DNA replication
 G2/M transition

Bibliography terms

Cancer
 Colon cancer
 Rectal cancer
 Tumor
 Gastrointestinal tumor
 Tumor suppressor
 Oncogene
 Carcinoma
 Sarcoma
 Adenoma
 Polyposis
 Polyps
 Cowden
 Inflammatory bowel disease
 Crohn
 Osteosarcoma
 Glioma
 Melanoma
 Leukemia
 Lymphoma