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| **Supplementary Table 1**. Some of the genes involved in tumorigenesis, which are targeted in commercially available sequencing kits | | | | | |
| **Gene** | **Location** | **Full name** | **UniGene** | **Function** | |
|  |  |  | **Native** | **When mutated** |
| ABL1 | 9q34.1 | c-abl oncogene 1, non-receptor tyrosine kinase | Hs.431048 | Cell growth, survival, adhesion, differentiation, DNA damage response, apoptosis | Chronic myeloid leukemia (CML), when translocated with BCR - also found in AML and ALL |
| APC | 5q21-q22 | adenomatous polyposis coli | Hs.158932 | Tumor suppressor, controls cell division | Colorectal cancer, Familial adenomatous polyposis |
| ATM | 11q22.3 | ataxia telangiectasia mutated | Hs.367437 | Cell division, DNA repair, normal development of nervous and immune system | When homozygous, ataxia-telangiectasia. When heterozygous, breast cancer and others |
| BARD1 | 2q34-q35 | BRCA1 associated RING domain 1 | Hs.591642 | Cell growth and division, together with BRCA1 | Uncertain risk in breast cancer and neuroblastoma |
| BRAF | 7q34 | v-raf murine sarcoma viral oncogene homolog B1 | Hs.550061 | In RAS/MAPK pathway, role in differentiation, migration, apoptosis | Cardiofaciocutaneous and multiple lentigines syndromes. Oncogene, somatic mutations associated with many cancers |
| BRCA1 | 17q21 | breast cancer 1, early onset | Hs.194143 | Tumor suppressor, gene regulation | Breast cancer, ovarian cancer, pancreatic cancer |
| BRCA2 | 13q12.3 | breast cancer 2, early onset | Hs.34012 | Tumor suppressor, gene regulation | Breast cancer, ovarian, pancreatic, prostate. Also Fanconi anemia type D1 when homozygous |
| BRIP1 | 17q22.2 | BRCA1 interacting protein C-terminal helicase 1 | Hs.128903 | DNA damage repair | Breast cancer when inherited heterozygous. Fanconi anemia when homozygous |
| CDH1 | 16q22.1 | cadherin 1, type 1, E-cadherin (epithelial) | Hs.461086 | Cell adhesion, cell signaling | Breast cancer, hereditary diffuse gastric cancer |
| CHEK2 | 22q12.1 | checkpoint kinase 2 | Hs.291363 | Tumor suppressor, detection of DNA damage and strand breaks | Breast cancer, Li-Fraumeni syndrome, other cancers |
| CTNNB1 | 3p21 | catenin (cadherin-associated protein), beta 1 | Hs.476018 | Cell adhesion, cell signaling | Pilomatricoma; colorectal, liver, ovarian cancer, medulloblastoma; desmoid fibromatosis |
| EPCAM | 2p21 | epithelial cell adhesion molecule | Hs.542050 | Calcium independent cell adhesion molecule in normal epithelium and gastrointestinal carcinoma | Hereditary nonpolyposis colorectal cancer, congenital tufting enteropathy |
| ERBB2 | 17q12 | Her-2/neu, v-erb-b2 erythroblastic leukemia viral oncogene homolog 2 | Hs.446352 | Encodes for growth factor receptors | Amplification found breast cancer, overexpression in receptors leads to aggressive forms of cancer |
| FGFR1 | 8p12 | fibroblast growth factor receptor 1 | Hs.264887 | Cell division, regulation of cell growth and maturation | Alterations found in cancers via proliferation, migration, angiogenesis |
| HRAS | 11p15.5 | v-Ha-ras Harvey rat sarcoma viral oncogene homolog | Hs.37003 | Regulates cell division through signal transduction | Oncogenes; involved in bladder, thyroid, kidney cancer; Costello syndrome |
| JAK2 | 9p24 | Janus kinase 2 | Hs.656213 | Cell growth and proliferation, role in hematopoiesis | Leukemia, essential thrombocythemia, primary myelofibrosis |
| KRAS | 12p12.1 | v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog | Hs.505033 | GTPase involved in cell division and differentiation | Oncogenes; involved in pancreatic, lung, and colorectal cancers; cardiofaciocutaneous and Noonan syndromes |
| MLH1 | 3p21.3 | mutL homolog 1, colon cancer, nonpolyposis type 2 | Hs.195364 | DNA damage repair | Lynch syndrome; when homozygous, also causes leukemia and neurofibromatosis |
| MRE11A | 11q21 | MRE11 meiotic recombination 11 homolog A | Hs.192649 | DNA double-strand break repair, telomere length maintenance, homologous recombination via exo- and endonuclease activity | Ataxia telangiectasia-like disorder, blocks meiotic recombination |
| MSH2 | 2p21 | mutS homolog 2, colon cancer, nonpolyposis type 1 | Hs.597656 | DNA damage repair | Lynch syndrome; endometrium, stomach, intenstine, liver cancer, etc. Homozygous mutations can also cause leukemia or lymphoma |
| MSH3 | 5q11-q12 | mutS homolog 3 | Hs.280987 | Heterodimer with MSH2, involved in post-replicative DNA mismatch repair system | Endometrial cancer |
| MSH6 | 2p16 | mutS homolog 6 | Hs.445052 | Post-replicative DNA damage repair | Lynch syndrome; endometrium, stomach, intenstine, liver cancer, etc. Homozygous mutations can also cause leukemia or lymphoma |
| MUTYH | 1p34.1 | mutY homolog | Hs.271353 | DNA damage repair by MYH glycosylase | Familial adenomatous polyposis |
| NRAS | 1p13.2 | neuroblastoma RAS viral (v-ras) oncogene homolog | Hs.486502 | Cell division and differentiation | Noonan syndrome, melanoma, other types |
| NBN | 8q21 | nibrin | Hs.492208 | DNA damage repair | Breast, ovarian, prostate cancer, melanoma, leukemia, Nijmegen breakage syndrome |
| PALB2 | 16p12.2 | partner and localizer of BRCA2 | Hs.444664 | Interacts with BRCA2; tumor suppressor | Breast cancer when inherited heterozygous. Fanconi anemia when homozygous |
| PIK3CA | 3q26.3 | phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit alpha | Hs.553498 | Role in signaling cascades involved in cell growth, survival, proliferation, motility and morphology | Oncogene, in breast, colorectal, ovarian, liver, gastric, lung cancers |
| PTEN | 10q23.3 | phosphatase and tensin homolog | Hs.500466 | Tumor suppressor, involved in apoptosis | Somatic mutations in prostate and endometrial cancer, glioblastoma, astrocytoma, melanoma |
| RAD50 | 5q31 | RAD50 homolog | Hs.633509 | DNA damage repair by holding the broken ends together during process | Suggested to contribute to breast cancer |
| RAD51C | 17q25.1 | RAD51 homolog C | Hs.412587 | DNA damage repair and meiotic homologous recombination | Hereditary breast and ovarian cancer, Fanconi anemia |
| SMAD4 | 18q21.1 | SMAD family member 4 | Hs.75862 | Controls gene activity and regulates cell proliferation | Colon, pancreas cancer; hereditary hemorrhagic telangiectasia, juvenile polyposis syndrome |
| SRC | 20q12-q13 | v-src sarcoma (Schmidt-Ruppin A-2) viral oncogene homolog | Hs.195659 | Proto-oncogene with role in regulation of embryonic development and cell growth | Increased activity in colon carcinoma cells |
| STK11 | 19p13.3 | serine/threonine kinase 11 | Hs.515005 | Tumor suppressor, role in tissue polarization and apoptosis | Breast cancer, non-small cell lung carcinoma, melanoma, pancreatic cancer; Peutz-Jeghers syndrome |
| TP53 | 17p13.1 | tumor protein p53 | Hs.437460 | Tumor suppressor, "guardian of the genome" (Lane, 1992) | Breast, bladder, colorectal cancer, osteosarcoma, Li-Fraumeni syndrome |