AVENIO ctDNA and Tumor Tissue Expanded Panel

This 77-gene panel, included in the AVENIO ctDNA Expanded Kit and the AVENIO Tumor Tissue Expanded Kit, contains biomarkers associated with various cancer types from 17 genes in the U.S. National Comprehensive Cancer (NCCN) Guidelines, plus 60 additional biomarkers relevant in clinical research.*

Gene	All Coding Regions	CRC	Lung	Breast	Gastric	Prostate	Bladder	Glioma	Melanoma	Ovarian	Thyroid	Pancreatic	Other Cancers	SNV	Indel‡	Fusion§	CNV§
ABL1													CML, ALL	●			
AKT1		●	●	●						●				●			
AKT2		●	●	●				●		●		●	Thymoma	●			
ALK			●										ALCL, Neuroblastoma, Inflammatory Myofibroblastic Tumor, Spitzoid Tumor, Large B-Cell Lymphoma, Rhabomyosarcoma	●	●	●	
APC		●			●			●				●	HCC, Medulloblastoma, Desmoid	●	●		
AR	●			●		●								●			
ARAF			●											●			
BRAF		●	●					●	●	●	●		Borderline Ovarian, Cholangiocarcinoma, Spitzoid Tumor, Pancreas Acinar Carcinoma, Melanocytic Nevus, GIST	●	●		
BRCA1	●			●						●		●		●			
BRCA2	●			●				●		●		●		●			
CCND1	●	●		●									CLL, B-ALL, MM, Mantle Cell Lymphoma	●			
CCND2	●												NHL, CLL	●			
CCND3	●						●	●					MM, Burkitt Lymphoma, other B-Cell Lymphomas	●			
CD274	●												Primary Mediastinal B-Cell Lymphoma, Hodgkin Lymphoma, CML, RCC, Lymphoepithelial-like Carcinoma	●			
CDK4	●			●					●				Liposarcoma	●			
CDK6				●				●					Endometrial Squamous Cancer, T-Cell Leukemia	●			
CDKN2A	●								●			●	Astrocytoma	●			
CSF1R				●		●				●			Endometrial, Testicular GCT, Malignant Histiocytosis	●			
CTNNB1		●								●			HCC, Medulloblastoma, Mesothelioma	●	●		

All coding regions are based on the longest transcript from Ensembl build 82.

* National Comprehensive Cancer Network. http://www.nccn.org. October 15, 2016.

‡ Indels are limited to variants in a pre-specified list of positions, referred to as "Loci of Interest", except for EGFR exon 19 long deletions, EGFR exon 20 long insertions and MET long insertions, which are not restricted to a pre-defined set of Indels.

§ Detection of Fusions and CNVs are limited to variants in a pre-specified list of positions, referred to as "Loci of Interest" in the AVENIO analysis software.

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DDR2			●										Lung Adenoid Cystic Carcinoma	●			
DPYD**		●												●			
EGFR	●		●					●						●	●		●
ERBB2	●	●†	●	●	●			●		●				●	●		●
ESR1	●			●										●			
EZH2													MDS, Mantle Cell Lymphoma, CML, DLBCL, Cholangiolocellular Carcinoma	●			
FBXW7	●	●		●									Endometrial, T-ALL, Lymphoblastic Leukemia	●			
FGFR1			●	●									MPN, NHL, Salivary Adenoma	●			
FGFR2			●	●	●								Endometrial	●		●	
FGFR3			●				●						MM, T-Cell Lymphoma, Cervical, Testicular GCT	●		●	
FLT1													CNS Hemangiomas	●			
FLT3													AML, ALL	●			
FLT4													Soft Tissue Sarcoma	●			
GATA3				●									RCC	●			
GNA11									●					●			
GNAQ									●					●			
GNAS		●											Pituitary Adenoma	●			
IDH1								●					AML	●			
IDH2								●					AML	●			
JAK2													ALL, AML, MPN, CML	●			
JAK3													Acute Megakaryocytic Leukemia, ETP ALL	●			
KDR			●										Angiosarcoma	●			
KEAP1	●		●											●			
KIT									●				AML, Testicular Germ Cell Tumors, GIST, Thymic Carcinoma	●	●		

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** Inactivating mutations are associated with sensitivity to fluoropyrimidines.

† Kavuri et al. Cancer Discovery 8: 832-41 (2015). PMID: 26243863.

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KRAS	●	●	●		●					●	●	●	AML, Juvenile Myelomonocytic Leukemia	●			
MAP2K1		●	●						●					●			
MAP2K2			●						●					●			
MET	●		●										Head and Neck Squamous Cell, HCC, Papillary RCC	●	●		●
MLH1	●	●		●	●					●			Endometrial, Small Intestine	●			
MSH2	●	●		●	●					●			Endometrial, Small Intestine	●			
MSH6	●	●		●	●					●			Endometrial, Small Intestine	●			
MTOR								●					Endometrial, Head and Neck SCC, Clear Cell RCC	●			
NF2	●												Meningioma, Acoustic Neuroma, Mesothelioma	●			
NFE2L2			●										Head and Neck SCC	●			
NRAS		●	●						●		●		MM, AML, Juvenile Myelomonocytic Leukemia	●			
NTRK1			●								●		Spitzoid Tumor	●		●	
PDCD1LG2	●												PMBL, Hodgkin Lymphoma	●			
PDGFRA													GIST, Hypereosinophilic Syndrome	●			
PDGFRB													MPN, AML, CMML, CML, Juvenile Myelomonocytic Leukemia	●			
PIK3CA		●		●	●			●		●	●		HCC	●	●		
PIK3R1		●						●		●				●			
PMS2	●	●		●	●					●			Endometrial, Small Intestine	●			
PTCH1													Basal Cell Carcinoma, Medulloblastoma	●			
PTEN	●	●		●		●		●			●		Endometrial, Head and Neck SCC, RCC	●	●		
RAF1						●							Pilocytic Astrocytoma	●			
RB1	●			●			●						Retinoblastoma, Small Cell Lung Carcinoma, Osteosarcoma	●			

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RET		●	●								●		Pheochromocytoma, Spitzoid Tumor, Mucosal Neuroma	●		●	
RNF43		●								●		●	Cholangiocarcinoma	●			
ROS1			●		●			●					Spitzoid Tumor, Cholangiocarcinoma, Borderline Ovarian	●		●	
SMAD4	●	●										●	Small Intestine	●			
SMO	●												Basal Cell Carcinoma, Medulloblastoma	●			
STK11	●		●						●			●	Testicular GCT	●			
TERT promoter							●	●	●				HCC, Basal Cell Carcinoma, Skin Squamous Cell, Mesothelioma, Medulloblastoma	●			
TP53	●	●	●	●	●			●		●		●	Various Leukemias, Adenocortical, Basal Cell Carcinoma, Esophageal, Soft Tissue and Bone Sarcomas, Head and Neck SCC	●	●§§		
TSC1							●						RCC, Angiomyolipoma	●	●		
TSC2													RCC, Angiomyolipoma, Head and Neck SCC	●			
UGT1A1††		●												●			
VHL	●												Hemangioblastoma, Pheochromocytoma, RCC	●			

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†† Certain variants are associated with sensitivity to irinotecan.
‡ Indels are limited to variants in a pre-specified list of positions, referred to as "Loci of Interest", except for EGFR exon 19 long deletions, EGFR exon 20 long insertions and MET long insertions, which are not restricted to a pre-defined set of Indels.
§ Detection of Fusions and CNVs are limited to variants in a pre-specified list of positions, referred to as "Loci of Interest" in the AVENIO analysis software.
§§ Indel for TP53 is called for tissue only, not plasma.

Associations of variations or mutations in these genes with the listed diseases were sourced from the following: COSMIC, GeneCards, My Cancer Genome. Analysis performed 11/2016.

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