

# SOMATIC MUTATIONS

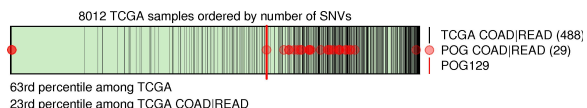
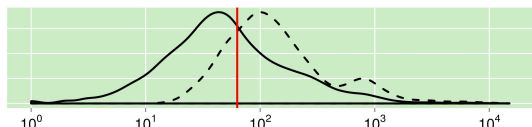
This section provides information for the somatic mutations described in the DETAILED GENOMIC ANALYSIS section, including SNVs and INDELS. Mutational burden analysis and mutation signature results are also reported in this section. Look in this section for details about mutation position, the sample in which mutations are detected, copy number and expression metrics.

## SMALL SOMATIC MUTATIONS

### SUMMARY OF SOMATIC EVENTS

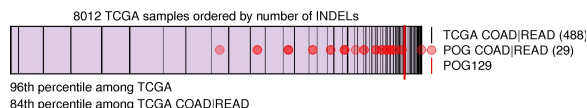
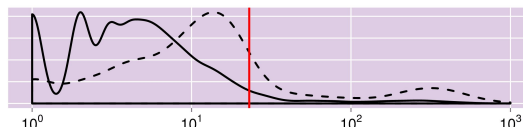
# Non-synonymous protein coding SNVs [Truncating]

63 [3]



# Protein coding indels [Frameshift]

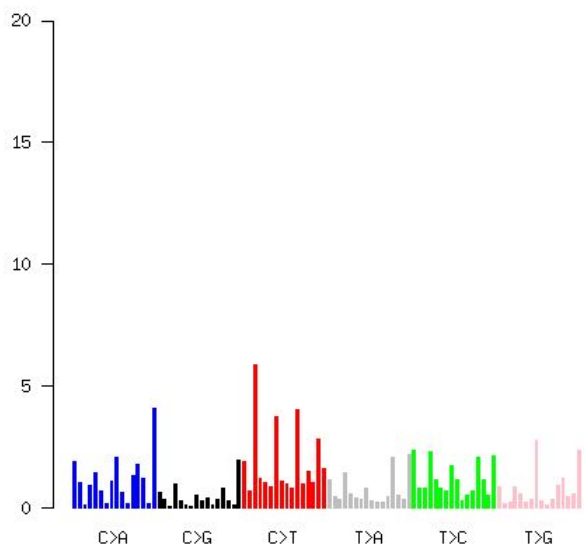
23 [18]



### MUTATIONAL SPECTRUM

Signature	Pearson	NNLS	Probable Associations	Additional Features	# Cancer types	Significant Cancer Type
Signature 1	0.497	0.188	Age	I	40	all
Signature 2	0.189	0.020	APOBEC	-	22	cervical cancer, urinary bladder cancer
Signature 3	0.077	0.000	BRCA1/2 mutations	I	3	breast cancer, ovarian cancer, pancreatic cancer
Signature 4	-0.024	0.000	Smoking	D,T	6	head and neck cancer, liver cancer, lung cancer, esophageal cancer
Signature 5	0.370	0.000	-	-	40	all
Signature 6	0.378	0.000	DNA MMR deficiency	I	17	colorectal cancer, uterine cancer
Signature 7	0.100	0.000	UV light	D,T	2	skin cancer, oral squamous cell carcinoma
Signature 8	0.286	0.191	-	-	2	breast cancer, medulloblastoma
Signature 9	0.640	0.284	IG gene hypermutation	-	2	chronic lymphocytic leukemia, diffuse large B-cell lymphoma
Signature 10	0.385	0.026	Pol e mutations	T	6	colorectal cancer, uterine cancer
Signature 11	0.023	0.000	Temozolomide	-	2	melanoma, glioblastoma
Signature 12	0.099	0.000	-	-	1	liver cancer
Signature 13	0.122	0.037	APOBEC	-	22	cervical cancer, urinary bladder cancer
Signature 14	0.290	0.000	-	-	2	uterine cancer, brain glioma
Signature 15	0.243	0.021	DNA MMR deficiency	I	2	stomach cancer, lung cancer
Signature 16	0.303	0.017	-	-	1	liver cancer
Signature 17	0.673	0.180	-	-	7	esophageal cancer, breast cancer, liver cancer, lung adenocarcinoma, B-cell lymphoma, stomach cancer, melanoma
Signature 18	0.211	0.020	-	-	3	neuroblastoma, breast cancer, stomach cancer
Signature 19	0.129	0.000	-	-	1	pilocytic astrocytoma
Signature 20	0.099	0.000	DNA MMR deficiency	I	2	stomach cancer, breast cancer
Signature 21	0.037	0.000	-	-	1	stomach cancer
Signature 22	-0.105	0.000	aristolochic acid	-	2	urinary bladder cancer, liver cancer
Signature 23	-0.022	0.000	-	-	1	liver cancer
Signature 24	-0.006	0.000	aflatoxin	-	1	liver cancer
Signature 25	0.186	0.000	-	-	1	Hodgkin's lymphoma cell lines
Signature 26	0.128	0.000	DNA MMR deficiency	I	4	breast cancer, cervical cancer, stomach cancer, uterine cancer
Signature 27	-0.042	0.001	-	I	1	renal clear cell carcinoma
Signature 28	0.451	0.013	-	-	1	stomach cancer
Signature 29	0.183	0.000	Tobacco chewing	-	1	oral squamous cell carcinoma
Signature 30	0.102	0.000	-	-	1	breast cancer

**SMALL SOMATIC MUTATIONS**



**SMALL MUTATIONS: GENOMIC DETAILS**

**VARIANTS OF POTENTIAL CLINICAL RELEVANCE**

Gene	Transcript	Protein Change	Location	Ref/Alt	Zygoty	Copy Change	LOH State	Ref/Alt DNA	Ref/Alt RNA	Expression (RPKM)	Fold Change vs. unknown	TCGA %ile
AKT1	ENST00000349310	p.Q79K	14:105243048	G>T	hom [2/2]	0	NLOH	27/14	313/700	na	na	na
APC	ENST00000457016	p.T1556fs	5:112175951	G>GA	na	+1	HET	na/21	na/32	3.71	-1.01	95
BRAF	ENST00000288602	p.V600E	7:140453136	A>T	het [2/3]	+1	HET	79/25	10/17	2.84	-1.36	90
SMAD4	ENST00000342988	p.D493N	18:48604655	G>A	hom [2/2]	0	NLOH	57/22	110/151	6.38	-1.55	79
	ENST00000588745	p.D397N	18:48604655	G>A	hom [2/2]	0	NLOH	57/22	110/151	6.38	-1.55	79

**VARIANTS OF PROGNOSTIC OR DIAGNOSTIC RELEVANCE**

Gene	Transcript	Protein Change	Location	Ref/Alt	Zygoty	Copy Change	LOH State	Ref/Alt DNA	Ref/Alt RNA	Expression (RPKM)	Fold Change vs. unknown	TCGA %ile
AKT1	ENST00000349310	p.Q79K	14:105243048	G>T	hom [2/2]	+2	NLOH	27/14	313/700	na	na	na
APC	ENST00000457016	p.T1556fs	5:112175951	G>GA	na	-1	HET	na/21	na/32	3.71	-1.01	95
BRAF	ENST00000288602	p.V600E	7:140453136	A>T	het [2/3]	+1	HET	79/25	10/17	2.84	-1.36	90
SMAD4	ENST00000342988	p.D493N	18:48604655	G>A	hom [2/2]	0	NLOH	57/22	110/151	6.38	-1.55	79
	ENST00000588745	p.D397N	18:48604655	G>A	hom [2/2]	-1	NLOH	57/22	110/151	6.38	-1.55	79

**VARIANTS OF BIOLOGICAL RELEVANCE**

Gene	Transcript	Protein Change	Location	Ref/Alt	Zygoty	Copy Change	LOH State	Ref/Alt DNA	Ref/Alt RNA	Expression (RPKM)	Fold Change vs. unknown	TCGA %ile
AKT1	ENST00000349310	p.Q79K	14:105243048	G>T	hom [2/2]	+1	NLOH	27/14	313/700	na	na	na
APC	ENST00000457016	p.T1556fs	5:112175951	G>GA	na	-1	HET	na/21	na/32	3.71	-1.01	95
BRAF	ENST00000288602	p.V600E	7:140453136	A>T	het [2/3]	-2	HET	79/25	10/17	2.84	-1.36	90
SMAD4	ENST00000342988	p.D493N	18:48604655	G>A	hom [2/2]	+2	NLOH	57/22	110/151	6.38	-1.55	79
	ENST00000588745	p.D397N	18:48604655	G>A	hom [2/2]	0	NLOH	57/22	110/151	6.38	-1.55	79

**VARIANTS OF UNKNOWN SIGNIFICANCE IN KNOWN CANCER-RELATED GENES**

Gene	Transcript	Protein Change	Location	Ref/Alt	Zygoty	Copy Change	LOH State	Ref/Alt DNA	Ref/Alt RNA	Expression (RPKM)	Fold Change vs. unknown	TCGA %ile
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## SMALL SOMATIC MUTATIONS

AKT1	ENST00000349310	p.Q79K	14:10524304 8	G>T	hom [2/2]	0	NLOH	27/14	313/700	na	na	na
APC	ENST00000457016	p.T1556fs	5:112175951	G>GA	na	+1	HET	na/21	na/32	3.71	-1.01	95
BRAF	ENST00000288602	p.V600E	7:140453136	A>T	het [2/3]	+1	HET	79/25	10/17	2.84	-1.36	90
SMAD4	ENST00000342988	p.D493N	18:48604655	G>A	hom [2/2]	0	NLOH	57/22	110/151	6.38	-1.55	79
	ENST00000588745	p.D397N	18:48604655	G>A	hom [2/2]	0	NLOH	57/22	110/151	6.38	-1.55	79