MIRACUM-Galaxy Gene Panel Analysereport

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Galaxy-Version

20.09

Mutationsanalyse

Zusammenfassung der identifizierten Mutationen

Dataset: Mutation Summary

Contents:

Mutationstyp	Genotyp	Anzahl	in_Tumorsuppressoren	in_Onkogenen	in_Hotspots
indel	heterozygot	134	17	7	0
snp	heterozygot	105	9	16	2
snp	homozygot	2	0	0	0

Identifizierte Mutationen in bekannten Krebsgenen

Dataset: Variants report for known cancer genes

Contents:

Gen	AA-Austausch	Funktion	Tumor-VAF (Coverage)	MAF	Cosmic	OG	TSG	Hotspot
ABL1	p.R535C	missense_variant	0.24 (874/3540)	0.0	.	1	0	.
ABL1	p.R804fs	frameshift_variant	0.26 (1052/4029)	.	.	1	0	.
APC	p.S1465fs	frameshift_variant	0.21 (224/1081)	.	COSM18873, COSM1432411, COSM1432412, COSM29573, COSM328654, COSM19688, COSM19694, COSM18838, COSM19332, COSM18983, COSM18931, COSM19141, COSM13864	0	1	.
ARAF	p.S388A	missense_variant	0.24 (1035/4207)	.	.	1	0	.
ATM	.	splice_region_variant	0.18 (53/179)	0.02	.	0	1	.
ATM	.	splice_region_variant	0.15 (42/168)	.	.	0	1	.
ATM	.	splice_region_variant	0.13 (42/167)	.	.	0	1	.
ATM	.	splice_region_variant	0.23 (74/199)	.	.	0	1	.

ATM	.	splice_region_variant	0.17 (70/251)	0.0	.	0	1	.
ATM	.	splice_region_variant	0.11 (71/628)	0.0	.	0	1	.
ATM	p.E2039K	missense_variant	0.19 (191/1009)	0.0	COSM1561120, COSM200671	0	1	.
AXIN1	p.P385S	missense_variant	0.26 (749/2830)	.	.	0	1	.
BCL2	p.R129S	missense_variant	0.54 (2981/4430)	0.0	.	1	0	.
BCR	p.A1204G	missense_variant	0.11 (527/4724)	0.04	.	1	0	.
BRAF	.	splice_region_variant	0.22 (75/348)	0.03	.	1	0	.
BRAF	p.V600E	missense_variant	0.13 (174/1365)	0.0	COSM18443, COSM6137, COSM476	1	0	0.0
CCND1	.	splice_region_variant	0.25 (563/2244)	.	.	1	0	.
CDKN2A	p.R139Q	missense_variant	0.27 (490/1825)	0.0	COSM216134, COSM13299, COSM238570, COSM1554789, COSM216135, COSM13613, COSM1624861, COSM13488, COSM238569	0	1	0.0
FLCN	p.H429fs	frameshift_variant	0.18 (339/1917)	0.01	COSM1381204, COSM1733577	0	1	.
GLI1	p.P755H	missense_variant	0.24 (715/2898)	.	.	1	0	.
GLI1	p.P765H	missense_variant	0.24 (709/2856)	.	.	1	0	.
GNAS	p.R1032H	missense_variant	0.11 (348/2981)	0.0	.	1	0	.
LMO1	p.S92T	missense_variant	0.23 (852/3677)	.	.	1	0	.
LYN	p.E110fs	frameshift_variant	0.14 (144/998)	0.0	.	1	0	.
MLH1	p.V185A	missense_variant	0.18 (145/772)	.	.	0	1	.
MLLT3	p.A252T	missense_variant	0.19 (421/2130)	.	.	1	0	.
MN1	p.Q533del	disruptive_inframe_deletion	0.15 (496/3279)	0.1	.	0	1	.
MSH2	p.A499T	missense_variant	0.17 (167/952)	.	.	0	1	.
MYCN	p.P45fs	frameshift_variant	0.19 (695/3800)	0.0	COSM1400341	1	0	.
NKX3-1	p.R36W	missense_variant	0.31 (437/1393)	.	.	0	1	.
NPM1	p.E170D	missense_variant	0.17 (126/722)	.	.	1	0	.
NPM1	.	splice_region_variant	0.25 (139/381)	0.01	.	1	0	.
NTRK1	p.V354I	missense_variant	0.23 (1001/4197)	0.0	.	1	0	.

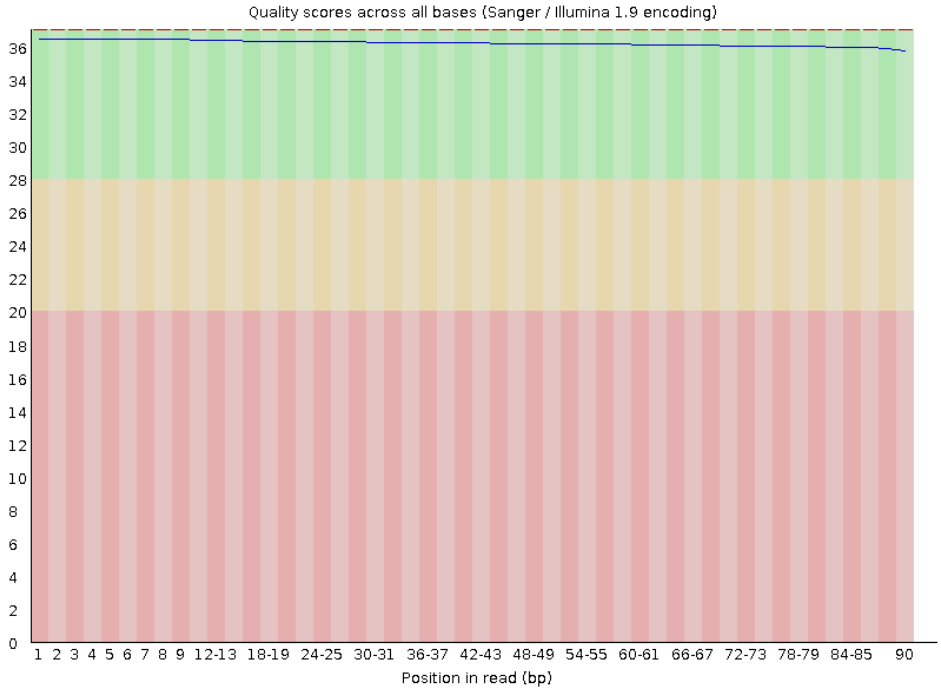
PAX5	p.P147H	missense_variant	0.21 (300/1441)	.	.	1	0	.
PBRM1	.	splice_region_variant	0.2 (113/408)	.	.	0	1	.
PBRM1	.	splice_region_variant	0.14 (76/371)	0.0	.	0	1	.
PMS2	.	splice_region_variant	0.11 (86/455)	0.0	.	0	1	.
PTCH1	p.Y1316fs	frameshift_variant	0.26 (1097/4337)	.	COSM1463794, COSM1463795, COSM1463796, COSM1463797	0	1	.
PTEN	.	splice_region_variant	0.31 (92/188)	0.0	.	0	1	.
PTEN	.	splice_region_variant	0.14 (41/137)	.	.	0	1	.
RET	p.R215C	missense_variant	0.23 (500/2186)	.	.	1	0	.
RET	.	splice_region_variant	0.22 (608/2757)	0.0	.	1	0	.
SDHA	.	splice_region_variant	0.27 (226/822)	0.01	.	0	1	.
SETD2	p.V2483I	missense_variant	0.21 (341/1578)	0.0	.	0	1	.
SRC	.	splice_donor_variant	0.23 (644/2705)	.	.	1	0	.
TFE3	p.G482fs	frameshift_variant	0.24 (1014/4314)	0.0	COSM1468292, COSM1468291, COSM1468293, COSM1468294	1	0	.
TSC1	.	splice_region_variant	0.13 (68/405)	.	.	0	1	.
TSC1	p.S334L	missense_variant	0.19 (234/1219)	0.0	.	0	1	.
TSC2	.	splice_region_variant	0.21 (522/2430)	0.0	.	0	1	.

Qualitaetskontrolle und Sequenzierungsstatistik

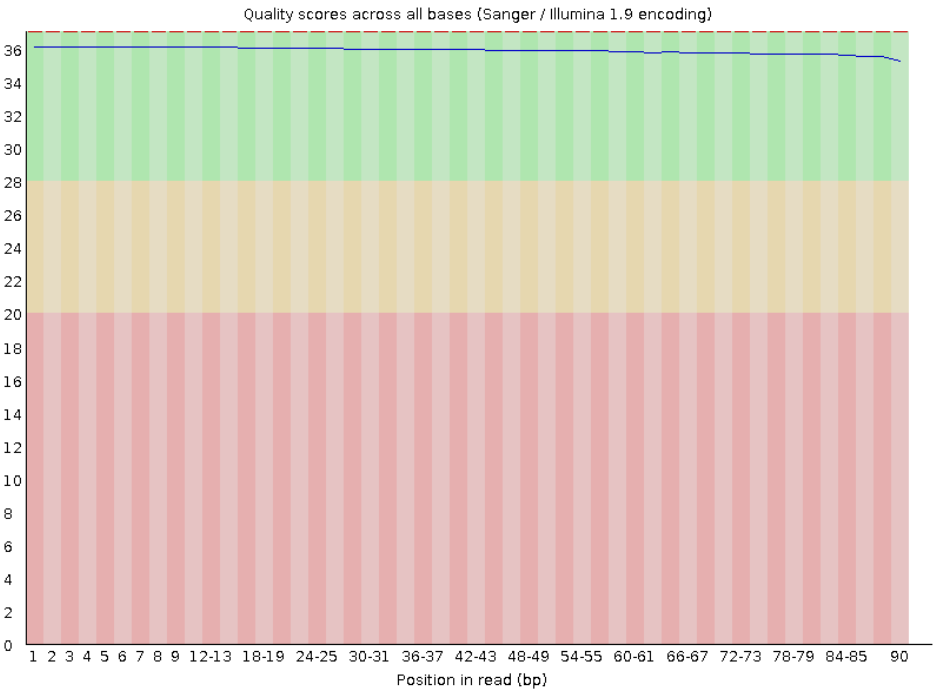
Qualitaet der Rohdaten

Die folgenden Abbildungen stellen die Basenqualitaeten für jede Position der sequenzierten Reads nach deren Trimming dar.

Forward Reads



Reverse Reads



Anhang A

Vollstaendige Variantenliste

Dataset: All variants report

Contents:

Gen	AA-Austausch	Funktion	VAF (Coverage)	MAF	Cosmic	OG	TSG	Hotspot
ABL1	p.R535C	missense_variant	0.24 (874/3540)	0.0	.	1	0	.
ABL1	p.R804fs	frameshift_variant	0.26 (1052/4029)	.	.	1	0	.
AC024560.3	.	splice_region_variant	0.37 (351/938)	.	.	0	0	.
AC027612.3	.	splice_region_variant	0.18 (42/234)	.	.	0	0	.
AC027612.3	.	splice_region_variant	0.16 (448/2846)	.	.	0	0	.
ANKRD26	.	splice_region_variant	0.18 (136/539)	.	.	0	0	.
APC	p.S1465fs	frameshift_variant	0.21 (224/1081)	.	COSM18873, COSM1432411, COSM1432412, COSM29573, COSM328654, COSM19688, COSM19694, COSM18838, COSM19332, COSM18983, COSM18931, COSM19141, COSM13864	0	1	.
AR	p.Q58L	missense_variant	0.16 (74/449)	0.0	COSM376477	0	0	.
ARAF	p.S388A	missense_variant	0.24 (1035/4207)	.	.	1	0	.
ARFRP1	p.R198M	missense_variant	0.2 (446/2252)	.	.	0	0	.
ARID1A	p.Q372	stop_gained	0.5 (128/257)	.	.	0	0	.
ARID1A	p.R1833H	missense_variant	0.2 (704/3511)	0.0	.	0	0	.
ARID1B	.	splice_region_variant	0.18 (217/1166)	0.0	.	0	0	.
ATM	.	splice_region_variant	0.18 (53/179)	0.02	.	0	1	.
ATM	.	splice_region_variant	0.15 (42/168)	.	.	0	1	.
ATM	.	splice_region_variant	0.13 (42/167)	.	.	0	1	.
ATM	.	splice_region_variant	0.23 (74/199)	.	.	0	1	.

ATM	.	splice_region_variant	0.17 (70/251)	0.0	.	0	1	.
ATM	.	splice_region_variant	0.11 (71/628)	0.0	.	0	1	.
ATM	p.E2039K	missense_variant	0.19 (191/1009)	0.0	COSM1561120, COSM200671	0	1	.
ATN1	p.Q498_Q502del	disruptive_inframe_deletion	0.36 (330/850)	.	COSM431781, COSM1476884, COSM1476883, COSM431782, COSM431783	0	0	.
ATR	.	splice_region_variant	0.3 (213/717)	0.01	.	0	0	.
ATR	p.I774fs	frameshift_variant	0.23 (161/690)	0.01	COSM1617015, COSM214499	0	0	.
ATRX	p.D975N	missense_variant	0.61 (340/552)	0.0	.	0	0	.
ATXN1	p.Q225dup	disruptive_inframe_insertion	0.21 (543/2569)	0.27	.	0	0	.
ATXN2	p.Q188del	disruptive_inframe_deletion	0.74 (565/727)	1.0	COSM404975	0	0	.
ATXN3	p.G315R	missense_variant	0.77 (287/359)	0.42	COSM318869	0	0	.
AXIN1	p.P385S	missense_variant	0.26 (749/2830)	.	.	0	1	.
BAGE2	.	splice_region_variant	0.74 (869/1153)	.	.	0	0	.
BAGE2	.	splice_region_variant	0.57 (385/664)	.	.	0	0	.
BCL2	p.R129S	missense_variant	0.54 (2981/4430)	0.0	.	1	0	.
BCOR	p.A570T	missense_variant	0.25 (1129/4535)	0.0	COSM1467919	0	0	.
BCR	p.A1204G	missense_variant	0.11 (527/4724)	0.04	.	1	0	.
BIVM-ERCC5	p.K597fs	frameshift_variant	0.16 (128/814)	.	.	0	0	.
BLM	p.N515fs	frameshift_variant	0.19 (179/935)	0.01	COSM1375442, COSM252959	0	0	.
BRAF	.	splice_region_variant	0.22 (75/348)	0.03	.	1	0	.
BRAF	p.V600E	missense_variant	0.13 (174/1365)	0.0	COSM18443, COSM6137, COSM476	1	0	0.0
C11orf30	p.T545A	missense_variant	0.22 (265/1209)	.	.	0	0	.
CCND1	.	splice_region_variant	0.25 (563/2244)	.	.	1	0	.
CD79A	p.P212fs	frameshift_variant	0.24 (720/3005)	0.0	COSM1284025, COSM189809	0	0	.
CDK8	p.F346fs	frameshift_variant	0.16 (153/937)	.	.	0	0	.

CDKN2A	p.R139Q	missense_variant	0.27 (490/1825)	0.0	COSM216134, COSM13299, COSM238570, COSM1554789, COSM216135, COSM13613, COSM1624861, COSM13488, COSM238569	0	1	0.0
CELF3	p.P355fs	frameshift_variant	0.22 (389/1840)	.	.	0	0	.
CHD2	p.V175fs	frameshift_variant	0.3 (386/1304)	0.0	COSM1375533, COSM1375534, COSM1375536, COSM1375535	0	0	.
CHD4	p.L931fs	frameshift_variant	0.17 (281/1640)	.	.	0	0	.
CHD4	p.K192E	missense_variant	0.22 (587/2621)	.	.	0	0	.
CHEK2P2	.	splice_region_variant	0.99 (1386/1387)	.	.	0	0	.
CREBBP	.	splice_region_variant	0.16 (111/535)	0.0	.	0	0	.
CREBBP	p.P543fs	frameshift_variant	0.19 (226/1189)	.	.	0	0	.
CSF3R	p.S469fs	frameshift_variant	0.2 (493/2431)	0.0	COSM1342335, COSM1342336	0	0	.
CTNNB1	.	splice_region_variant	0.18 (36/203)	0.01	.	0	0	.
CUL3	p.A371V	missense_variant	0.21 (240/1146)	.	.	0	0	.
CUX1	p.R447C	missense_variant	0.17 (610/3558)	0.0	.	0	0	.
DCP1B	p.Q261del	disruptive_inframe_deletion	0.1 (45/432)	0.04	.	0	0	.
DENND4B	p.Q908_Q910del	conservative_inframe_deletion	0.21 (321/1453)	0.46	COSM1319935, COSM1319934	0	0	.
DICER1	p.E1420dup	disruptive_inframe_insertion	0.16 (276/1764)	.	.	0	0	.
DICER1	.	splice_region_variant	0.12 (69/568)	0.0	.	0	0	.
DNMT1	.	splice_region_variant	0.19 (292/1236)	0.0	.	0	0	.
DNMT3B	p.L454fs	frameshift_variant	0.23 (729/3161)	.	COSM1411124, COSM1411123	0	0	.
DOT1L	p.S1520L	missense_variant	0.21 (257/1169)	.	.	0	0	.
E2F4	p.S319del	disruptive_inframe_deletion	0.35 (705/1912)	0.33	COSM435515, COSM435516	0	0	.
EP300	.	splice_region_variant	0.29 (202/588)	0.0	.	0	0	.
EP400	p.Q2777_Q2778del	disruptive_inframe_deletion	0.1 (303/2654)	.	.	0	0	.
EPHA5	p.I670R	missense_variant	0.18 (162/908)	.	.	0	0	.
EPHA5	p.A662D	missense_variant	0.19 (164/848)	.	.	0	0	.
EPHA7	p.A409T	missense_variant	0.18 (299/1641)	.	.	0	0	.

ERBB4	.	splice_region_variant	0.18 (51/186)	0.0	.	0	0	.
ERCC2	.	splice_acceptor_variant	0.25 (854/3316)	.	.	0	0	.
ESR1	p.Q500dup	disruptive_inframe_insertion	0.18 (879/4842)	.	.	0	0	.
ETV5	.	splice_region_variant	0.2 (225/1092)	0.0	.	0	0	.
FANCD2	.	splice_region_variant	0.14 (132/930)	.	.	0	0	.
FANCE	p.P310fs	frameshift_variant	0.23 (563/2432)	0.0	.	0	0	.
FAS	.	splice_region_variant	0.18 (159/861)	0.0	.	0	0	.
FBXW7	.	splice_region_variant	0.22 (88/295)	.	.	0	0	.
FLCN	p.H429fs	frameshift_variant	0.18 (339/1917)	0.01	COSM1381204, COSM1733577	0	1	.
FLT1	p.W1083R	missense_variant	0.18 (214/1180)	0.0	.	0	0	.
FLT4	p.R1189H	missense_variant	0.21 (742/3462)	0.0	.	0	0	.
FOXP1	.	splice_region_variant	0.1 (49/247)	.	.	0	0	.
FOXP1	.	splice_region_variant	0.25 (121/319)	.	.	0	0	.
FOXP1	.	splice_region_variant	0.14 (67/265)	.	.	0	0	.
FOXP2	p.Q181del	disruptive_inframe_deletion	0.26 (209/816)	0.0	.	0	0	.
GATA1	p.T396fs	frameshift_variant	0.27 (686/2563)	.	.	0	0	.
GATA1	p.T402fs	frameshift_variant	0.29 (629/2260)	.	.	0	0	.
GATA2	p.G310D	missense_variant	0.25 (597/2334)	.	.	0	0	.
GLI1	p.P755H	missense_variant	0.24 (715/2898)	.	.	1	0	.
GLI1	p.P765H	missense_variant	0.24 (709/2856)	.	.	1	0	.
GNAQ	.	splice_region_variant	0.13 (91/510)	.	.	0	0	.
GNAS	p.R1032H	missense_variant	0.11 (348/2981)	0.0	.	1	0	.
HIST1H1E	p.A112V	missense_variant	0.49 (126/253)	0.0	.	0	0	.
HIST1H3E	p.R9C	missense_variant	0.22 (454/2043)	.	.	0	0	.
HIST2H2BE	p.S37fs	frameshift_variant	0.19 (507/2723)	.	.	0	0	.
HIST3H3	p.A48T	missense_variant	0.5 (1138/2270)	.	.	0	0	.
HIST3H3	p.R41H	missense_variant	0.24 (465/1897)	.	.	0	0	.
HNF1A	p.A251T	missense_variant	0.24 (1065/4346)	.	.	0	0	.

HNF1A	p.P291fs	frameshift_variant	0.22 (702/3246)	0.27	COSM935974, COSM1476243, COSM1684350, COSM1684349	0	0	.
HSP90AA1	p.K367E	missense_variant	0.13 (134/937)	0.0	.	0	0	.
HTT	p.Q36_Q37del	disruptive_inframe_deletion	0.13 (175/1242)	0.74	COSM1190754	0	0	.
IGF1R	.	splice_region_variant	0.21 (234/1141)	0.0	.	0	0	.
INHA	p.T212M	missense_variant	0.24 (971/4014)	0.0	.	0	0	.
KDM5A	p.G1200fs	frameshift_variant	0.43 (1144/2703)	0.0	COSM1180774, COSM1180773	0	0	.
KDM5A	p.P469S	missense_variant	0.16 (171/1035)	.	.	0	0	.
KDM5C	p.R1165C	missense_variant	0.22 (978/4343)	.	.	0	0	.
KDM6A	p.T733M	missense_variant	0.2 (412/2014)	0.0	.	0	0	.
KDM6A	.	splice_region_variant	0.19 (69/110)	0.0	.	0	0	.
KEL	p.R447Q	missense_variant	0.34 (1369/3918)	0.0	.	0	0	.
KIAA2018	p.Q1473del	disruptive_inframe_deletion	0.24 (167/585)	0.57	COSM1484461, COSM1684935	0	0	.
KMT2B	p.P1961fs	frameshift_variant	0.29 (846/2964)	.	.	0	0	.
KMT2C	.	splice_region_variant	0.41 (382/931)	0.0	COSM1721288, COSM1721289	0	0	.
KMT2C	p.S3567T	missense_variant	0.37 (992/2634)	.	.	0	0	.
KMT2C	.	splice_region_variant	0.17 (103/517)	0.0	.	0	0	.
LAMP1	.	splice_region_variant	0.15 (159/676)	0.01	.	0	0	.
LMO1	p.S92T	missense_variant	0.23 (852/3677)	.	.	1	0	.
LRP1B	.	splice_region_variant	0.2 (133/673)	0.0	.	0	0	.
LRP1B	.	splice_region_variant	0.28 (152/472)	.	.	0	0	.
LRP1B	p.D2085V	missense_variant	0.17 (174/1008)	.	.	0	0	.
LYN	p.E110fs	frameshift_variant	0.14 (144/998)	0.0	.	1	0	.
MAGI1	p.Q421dup	conservative_inframe_insertion	0.3 (868/2934)	0.48	COSM1485456, COSM1048140, COSM1048138	0	0	.
MAP3K1	.	splice_region_variant	0.21 (183/863)	.	.	0	0	.
MAP3K4	p.R1052H	missense_variant	0.18 (118/626)	.	.	0	0	.
MAP3K4	p.A1199dup	disruptive_inframe_insertion	0.1 (461/4495)	0.01	.	0	0	.
MED12	p.T582M	missense_variant	0.26 (386/1434)	0.0	.	0	0	.

MLH1	p.V185A	missense_variant	0.18 (145/772)	.	.	0	1	.
MLLT3	p.A252T	missense_variant	0.19 (421/2130)	.	.	1	0	.
MN1	p.Q533del	disruptive_inframe_deletion	0.15 (496/3279)	0.1	.	0	1	.
MRE11A	.	splice_region_variant	0.19 (115/517)	.	.	0	0	.
MRE11A	p.E95K	missense_variant	0.46 (408/875)	0.0	.	0	0	.
MSH2	p.A499T	missense_variant	0.17 (167/952)	.	.	0	1	.
MSH3	p.K383fs	frameshift_variant	0.35 (191/538)	0.0	COSM1438888, COSM1568178, COSM1642798	0	0	.
MSH6	p.R577H	missense_variant	0.14 (162/1162)	0.0	COSM1215570	0	0	.
MSH6	p.F1088fs	frameshift_variant	0.19 (329/1759)	0.0	COSM308681, COSM330655, COSM13395	0	0	.
MSH6	.	splice_region_variant	0.17 (114/461)	0.0	.	0	0	.
MSH6	.	splice_region_variant	0.13 (89/436)	0.0	.	0	0	.
MST1	p.I693R	missense_variant	0.1 (442/4100)	0.0	.	0	0	.
MST1P2	.	splice_acceptor_variant	0.15 (1123/7385)	.	.	0	0	.
MST1R	p.R504H	missense_variant	0.19 (616/3227)	0.0	.	0	0	.
MTOR	p.R2197C	missense_variant	0.17 (396/2268)	.	.	0	0	.
MTOR	p.T1780I	missense_variant	0.21 (711/3324)	.	.	0	0	.
MYCN	p.P45fs	frameshift_variant	0.19 (695/3800)	0.0	COSM1400341	1	0	.
NCOA3	p.Q1264del	disruptive_inframe_deletion	0.49 (1969/3977)	0.41	COSM128730	0	0	.
NCOA6	p.Q277del	disruptive_inframe_deletion	0.12 (165/1337)	0.03	.	0	0	.
NCOR1	p.A793T	missense_variant	0.19 (507/2553)	.	.	0	0	.
NCOR1	p.E529fs	frameshift_variant	0.19 (117/631)	.	.	0	0	.
NCOR1P2	.	splice_region_variant	0.51 (619/1197)	0.51	.	0	0	.
NCOR2	p.Q509_Q510del	disruptive_inframe_deletion	0.28 (477/1631)	.	.	0	0	.
NCOR2	p.Q510dup	conservative_inframe_insertion	0.13 (219/1688)	0.44	COSM1476308, COSM1476307	0	0	.
NF1P3	.	splice_region_variant	0.99 (418/418)	1.0	.	0	0	.
NF1P6	.	splice_donor_variant	0.39 (468/1166)	.	.	0	0	.
NF1P6	.	splice_region_variant	0.17 (155/901)	.	.	0	0	.

NKX3-1	p.R36W	missense_variant	0.31 (437/1393)	.	.	0	1	.
NOTCH1	p.R1586H	missense_variant	0.24 (1071/4450)	0.0	.	0	0	.
NOTCH1	p.A375T	missense_variant	0.23 (931/3960)	.	.	0	0	.
NOTCH2	p.N1999fs	frameshift_variant	0.13 (158/1222)	0.0	.	0	0	.
NPM1	p.E170D	missense_variant	0.17 (126/722)	.	.	1	0	.
NPM1	.	splice_region_variant	0.25 (139/381)	0.01	.	1	0	.
NTRK1	p.V354I	missense_variant	0.23 (1001/4197)	0.0	.	1	0	.
NTRK2	p.P382S	missense_variant	0.19 (148/763)	.	.	0	0	.
NUP93	.	splice_donor_variant	0.18 (367/2014)	0.0	.	0	0	.
PARP1	p.P359fs	frameshift_variant	0.25 (1005/4098)	.	.	0	0	.
PAX5	p.P147H	missense_variant	0.21 (300/1441)	.	.	1	0	.
PBRM1	.	splice_region_variant	0.2 (113/408)	.	.	0	1	.
PBRM1	.	splice_region_variant	0.14 (76/371)	0.0	.	0	1	.
PGR	p.A650V	missense_variant	0.21 (197/912)	.	.	0	0	.
PGR	p.P104A	missense_variant	0.17 (690/3949)	.	.	0	0	.
PIK3CD	p.A310V	missense_variant	0.23 (622/2690)	.	COSM1345058	0	0	.
PIK3R1	p.I82fs	frameshift_variant	0.14 (237/1668)	.	.	0	0	.
PIK3R1	p.K448fs	frameshift_variant	0.17 (94/559)	0.0	.	0	0	.
PLCG2	p.V411M	missense_variant	0.21 (675/3210)	.	.	0	0	.
PMS2	.	splice_region_variant	0.11 (86/455)	0.0	.	0	1	.
POLD1	p.R225H	missense_variant	0.31 (381/1215)	0.0	.	0	0	.
POLD1	.	splice_region_variant	0.19 (376/1370)	0.0	.	0	0	.
POLD1	.	splice_region_variant	0.24 (483/1477)	0.0	.	0	0	.
POLD1	p.R1035H	missense_variant	0.29 (771/2581)	0.0	.	0	0	.
POLE	.	splice_region_variant	0.12 (393/1995)	0.0	.	0	0	.
POLE	p.R114	stop_gained	0.2 (291/1459)	.	.	0	0	.
POLE	p.Y84C	missense_variant	0.24 (516/2113)	.	.	0	0	.

PTCH1	p.Y1316fs	frameshift_variant	0.26 (1097/4337)	.	COSM1463794, COSM1463795, COSM1463796, COSM1463797	0	1	.
PTEN	.	splice_region_variant	0.31 (92/188)	0.0	.	0	1	.
PTEN	.	splice_region_variant	0.14 (41/137)	.	.	0	1	.
PTPRD	.	splice_region_variant	0.14 (92/654)	0.71	.	0	0	.
PTPRD	p.P867S	missense_variant	0.14 (158/1078)	.	.	0	0	.
PTPRS	p.R1081C	missense_variant	0.25 (981/3929)	0.0	.	0	0	.
QKI	.	splice_region_variant	0.31 (194/478)	0.0	.	0	0	.
QKI	.	splice_region_variant	0.11 (70/354)	0.0	.	0	0	.
RAD21	.	splice_region_variant	0.13 (63/357)	0.0	.	0	0	.
RAD50	p.K722fs	frameshift_variant	0.21 (242/1137)	0.0	COSM1433045, COSM1433046	0	0	.
RAD51B	p.A149T	missense_variant	0.22 (281/1263)	0.0	.	0	0	.
RAD54L	p.A117T	missense_variant	0.2 (585/2835)	.	.	0	0	.
RAI1	p.Q290_Q291del	disruptive_inframe_deletion	0.12 (99/380)	0.06	COSM1190909	0	0	.
RAI1	p.Q291del	disruptive_inframe_deletion	0.55 (454/735)	0.29	COSM436135, COSM1479304	0	0	.
RANBP2	.	splice_region_variant	0.12 (115/925)	0.0	COSM1641610, COSM1641609	0	0	.
RANBP2	p.K2576fs	frameshift_variant	0.26 (101/392)	.	.	0	0	.
RBM10	p.A659G	missense_variant	0.22 (641/2876)	.	.	0	0	.
RET	p.R215C	missense_variant	0.23 (500/2186)	.	.	1	0	.
RET	.	splice_region_variant	0.22 (608/2757)	0.0	.	1	0	.
RFWD2	.	splice_region_variant	0.15 (70/360)	.	.	0	0	.
RGPD3	p.S937R	missense_variant	0.15 (136/875)	0.0	COSM440948, COSM440947	0	0	.
RGPD3	.	splice_region_variant	0.14 (110/753)	.	.	0	0	.
RGPD3	.	splice_region_variant	0.28 (78/277)	0.0	.	0	0	.
RGPD4	p.L518F	missense_variant	0.24 (260/1094)	0.01	COSM227222	0	0	.
RGPD4	.	splice_region_variant	0.11 (80/756)	0.0	.	0	0	.
RGPD8	p.E928K	missense_variant	0.17 (65/364)	0.05	COSM1318197, COSM1318196	0	0	.
RGPD8	.	splice_region_variant	0.25 (122/449)	0.0	.	0	0	.

RGPD8	.	splice_region_variant	0.22 (176/803)	0.0	.	0	0	.
RNF43	p.G659fs	frameshift_variant	0.45 (1874/4188)	0.0	COSM1384748, COSM270052	0	0	.
RPL14	p.A157_A159dup	conservative_inframe_insertion	0.2 (109/307)	0.58	COSM1044558, COSM1044560, COSM1044559	0	0	.
RPL14	p.A156_A159dup	conservative_inframe_insertion	0.29 (154/352)	.	.	0	0	.
RPL14	p.A155_A159dup	conservative_inframe_insertion	0.11 (56/254)	0.3	.	0	0	.
RPS6KB1	.	splice_region_variant	0.22 (183/841)	0.0	.	0	0	.
RUNX2	p.S99fs	frameshift_variant	0.29 (72/257)	.	COSM1444867, COSM1444868	0	0	.
SDHA	.	splice_region_variant	0.27 (226/822)	0.01	.	0	1	.
SETD2	p.V2483I	missense_variant	0.21 (341/1578)	0.0	.	0	1	.
SMAD3	p.A168T	missense_variant	0.22 (650/2770)	0.0	.	0	0	.
SMAD3	p.F314fs	frameshift_variant	0.22 (842/3852)	.	.	0	0	.
SMARCA2	p.Q236del	disruptive_inframe_deletion	0.27 (738/2630)	0.26	.	0	0	.
SMARCA4	p.E1144K	missense_variant	0.2 (734/3575)	.	.	0	0	.
SNCAIP	p.Q749	stop_gained	0.22 (815/3596)	.	.	0	0	.
SPEN	.	splice_region_variant	0.11 (64/493)	0.0	.	0	0	.
SPEN	p.R732	stop_gained	0.23 (919/3891)	.	.	0	0	.
SPTA1	.	splice_region_variant	0.3 (91/288)	.	.	0	0	.
SRC	.	splice_donor_variant	0.23 (644/2705)	.	.	1	0	.
STAT4	.	splice_region_variant	0.13 (78/594)	0.01	.	0	0	.
STAT5B	p.Q368fs	frameshift_variant	0.21 (564/2655)	0.0	COSM1383342, COSM1383343	0	0	.
SUZ12P	.	splice_region_variant	0.11 (32/203)	0.42	.	0	0	.
SUZ12P	.	splice_region_variant	0.27 (77/248)	.	.	0	0	.
TAF1	p.V499L	missense_variant	0.24 (221/925)	.	.	0	0	.
TAF1L	p.A852fs	frameshift_variant	0.21 (201/957)	.	COSM20659	0	0	.
TAF1L	p.K665fs	frameshift_variant	0.25 (382/1555)	0.0	COSM1462011, COSM20437, COSM20436	0	0	.
TBP	p.Q72dup	disruptive_inframe_insertion	0.23 (697/3022)	0.73	COSM247745	0	0	.
TET1	p.K22fs	frameshift_variant	0.27 (286/1082)	.	COSM1348704	0	0	.

TFE3	p.G482fs	frameshift_variant	0.24 (1014/4314)	0.0	COSM1468292, COSM1468291, COSM1468293, COSM1468294	1	0	.
TMPRSS2	p.D375V	missense_variant	0.19 (462/2442)	0.0	.	0	0	.
TOP2A	p.T1205fs	frameshift_variant	0.23 (399/1732)	0.0	COSM1382971	0	0	.
TPRXL	p.P69H	missense_variant	0.42 (208/467)	0.58	.	0	0	.
TSC1	.	splice_region_variant	0.13 (68/405)	.	.	0	1	.
TSC1	p.S334L	missense_variant	0.19 (234/1219)	0.0	.	0	1	.
TSC2	.	splice_region_variant	0.21 (522/2430)	0.0	.	0	1	.
TSPAN31	.	splice_region_variant	0.17 (156/667)	.	.	0	0	.
TSPAN31	.	splice_region_variant	0.18 (174/685)	.	.	0	0	.
XPO1	.	splice_region_variant	0.17 (62/291)	.	.	0	0	.
ZFHX3	.	splice_region_variant	0.14 (120/683)	.	.	0	0	.
ZFHX3	p.E763fs	frameshift_variant	0.3 (590/1961)	.	.	0	0	.

Gen-Informationen

Dataset: Affected genes details

Contents:

ABL1	
chrom	chr9
synonym	ABL,JTK7,c-ABL,p150
hgnc_id	76
entrez_id	25
rvis_pct	2.854446803
is_OG	1
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	chronic_myeloid_leukemia,resistant_to_imatinib| leukemia,philadelphia_chromosome-positive,resistant_to_imatinib
gene_civic_url	https://civic.genome.wustl.edu/links/genes/4

description	ABL1 is most relevant to cancer in its role in the BCR-ABL fusion protein that has become a signature of chronic myeloid leukemia (CML). Cells harboring this fusion have shown sensitivity to imatinib, greatly improving the prognostic outlook of the disease. However, additional mutations in ABL1 have been shown to confer resistance to imatinib. In these resistance cases, second-generation tyrosine kinase inhibitors such as dasatinib and nilotinib have exhibited some efficacy and are currently undergoing clinical trials for treating acquired resistance in CML.
---	---
AC024560.3	
chrom	chr3
synonym	.
hgnc_id	.
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
AC027612.3	
chrom	chr2
synonym	.
hgnc_id	.
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
AKT3	
chrom	chr1
synonym	PKBG,PRKBG,RAC-gamma
hgnc_id	393
entrez_id	10000
rvis_pct	35.42108988
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	bardet-biedl_syndrome| megalencephaly-polymicrogyria-polydactyly-hydrocephalus_syndrome_2| renal_dysplasia_and_retinal_aplasia
gene_civic_url	https://civic.genome.wustl.edu/links/genes/7936

description	.
---	---
ANKRD26	
chrom	chr10
synonym	THC2,KIAA1074
hgnc_id	29186
entrez_id	22852
rvis_pct	97.98301486
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	thrombocytopenia| thrombocytopenia_2
gene_civic_url	.
description	.
---	---
APC	
chrom	chr5
synonym	DP3,PPP1R46,DP2,DP2.5
hgnc_id	583
entrez_id	324
rvis_pct	0.902335456
is_OG	0
is_TS	1
in_cgi_biomarkers	1
clinvar_gene_phenotype	apc-associated_polyposis_disorders| adenomatous_colonic_polyposis| adenomatous_polyposis_coli,susceptibility_to| adenomatous_polyposis_coli_with_congenital_cholesteatoma| brain_tumor-polyposis_syndrome_2| breast_cancer,susceptibility_to| carcinoma_of_colon| colon_cancer| colorectal_adenoma| colorectal_cancer,susceptibility_to| desmoid_disease,hereditary| familial_adenomatous_polyposis_1| familial_cancer_of_breast| familial_colorectal_cancer| familial_multiple_polyposis_syndrome| gardner_syndrome| hepatoblastoma| hepatocellular_carcinoma| hereditary_cancer-predisposing_syndrome| monoclonal_b-cell_lymphocytosis| neoplasm_of_stomach| periampullary_adenoma
gene_civic_url	https://civic.genome.wustl.edu/links/genes/66
description	.
---	---
AR	
chrom	chrX
synonym	NR3C4,HUMARA,AIS,SBMA,SMAX1,DHTR
hgnc_id	644
entrez_id	367
rvis_pct	17.30950696
is_OG	0
is_TS	0
in_cgi_biomarkers	1

clinvar_gene_phenotype	androgen_insensitivity,partial,with_breast_cancer| androgen_resistance_syndrome| hypospadias_1,x-linked| malignant_tumor_of_prostate| prostate_cancer_susceptibility| reifenstein_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/67
description	.
---	---
ARAF	
chrom	chrX
synonym	ARAF1
hgnc_id	646
entrez_id	369
rvis_pct	22.35786742
is_OG	1
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/3
description	ARAF has recently become increasingly considered for its oncogenic potential. Its potential as a target for informing clinical action was demonstrated by a single case of advanced lung adenocarcinoma harboring an S214C mutation that, when treated with sorafenib, acheived near-complete clinical remission. This finding has brought new focus on ARAF as a marker that should be assayed for in cancer treatment.
---	---
ARFRP1	
chrom	chr20
synonym	ARP,ARL18,Arp1
hgnc_id	662
entrez_id	10139
rvis_pct	43.29440906
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	long_qt_syndrome
gene_civic_url	.
description	.
---	---
ARID1A	
chrom	chr1
synonym	BAF250a,P270,BAF250,C1orf4,SMARCF1,B120,C10rf4
hgnc_id	11110
entrez_id	8289
rvis_pct	0.743099788
is_OG	0
is_TS	0
in_cgi_biomarkers	1

clinvar_gene_phenotype	coffin_siris/intellectual_disability| inborn_genetic_diseases| mental_retardation,autosomal_dominant_14
gene_civic_url	https://civic.genome.wustl.edu/links/genes/6559
description	.
---	---
ARID1B	
chrom	chr6
synonym	BAF250b,p250R,DAN15,ELD/OSA1,KIAA1235,6A3-5
hgnc_id	18040
entrez_id	57492
rvis_pct	0.802075961
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	absent_speech| agenesis_of_corpus_callosum| bilateral_cryptorchidism| blepharophimosis| coffin-siris_syndrome| coffin-siris_syndrome_1| coffin_siris/ intellectual_disability| constipation| decreased_body_weight| delayed_speech_and_language_development| failure_to_thrive| global_developmental_delay| hypertrichosis| inborn_genetic_diseases| intellectual_disability| intellectual_disability,moderate| long_eyelashes| mental_retardation,autosomal_dominant_12| microcephaly| nail_dysplasia| neonatal_hypotonia| neurological_speech_impairment| recurrent_respiratory_infections| seizures| short_stature| thick_lower_lip_vermilion| thin_upper_lip_vermilion| dysmorphy| intellectual_deficiency
gene_civic_url	.
description	.
---	---
ATM	
chrom	chr11
synonym	TEL1,ATD,ATDC,ATC,ATA,TELO1
hgnc_id	795
entrez_id	472
rvis_pct	95.50601557
is_OG	0
is_TS	1
in_cgi_biomarkers	1
clinvar_gene_phenotype	ataxia| ataxia-telangiectasia,complementation_group_e| ataxia-telangiectasia_syndrome| ataxia-telangiectasia_variant| ataxia-telangiectasia_without_immunodeficiency| b-cell_non-hodgkin_lymphoma| bilateral_breast_carcinoma| breast_cancer,early-onset| breast_cancer,susceptibility_to| conjunctival_telangiectasia| hereditary_cancer-predisposing_syndrome| hereditary_cancer-predisposing_syndrome,not_specified| immunodeficiency| inborn_genetic_diseases| mantle_cell_lymphoma| oculomotor_apraxia| t-cell_prolymphocytic_leukemia
gene_civic_url	https://civic.genome.wustl.edu/links/genes/69
description	ATM is a DNA-damage response gene that is commonly mutated in cancer. Germline mutations in this gene are thought to contribute to breast cancer susceptibility, and PARP inhibition is currently being studied for it's potential in treating these patients.
---	---
ATN1	
chrom	chr12

synonym	DRPLA,B37,D12S755E
hgnc_id	3033
entrez_id	1822
rvis_pct	9.341825902
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
ATR	
chrom	chr3
synonym	MEC1,SCKL,SCKL1,FRP1
hgnc_id	882
entrez_id	545
rvis_pct	2.789573013
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	cutaneous_telangiectasia_and_cancer_syndrome,familial| hereditary_cancer-predisposing_syndrome| seckel_syndrome| seckel_syndrome_1
gene_civic_url	https://civic.genome.wustl.edu/links/genes/524
description	.
---	---
ATRX	
chrom	chrX
synonym	XH2,RAD54,JMS,XNP
hgnc_id	886
entrez_id	546
rvis_pct	9.754659118
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	atr-x_syndrome| atr-x_syndrome,not_specified| acquired_hemoglobin_h_disease| inborn_genetic_diseases| intellectual_disability| mental_retardation-hypotonic_facies_syndrome,x-linked| mental_retardation-hypotonic_facies_syndrome_x-linked,1
gene_civic_url	https://civic.genome.wustl.edu/links/genes/525
description	.
---	---
ATXN1	
chrom	chr6
synonym	SCA1,D6S504E,ATX1

hgnc_id	10548
entrez_id	6310
rvis_pct	20.93654164
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
ATXN2	
chrom	chr12
synonym	TNRC13,SCA2,ATX2
hgnc_id	10555
entrez_id	6311
rvis_pct	8.539749941
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
ATXN3	
chrom	chr14
synonym	MJD,ATX3,SCA3,JOS
hgnc_id	7106
entrez_id	4287
rvis_pct	71.26680821
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	azorean_disease
gene_civic_url	.
description	.
---	---
AXIN1	
chrom	chr16
synonym	PPP1R49
hgnc_id	903
entrez_id	8312
rvis_pct	4.016277424

is_OG	0
is_TS	1
in_cgi_biomarkers	.
clinvar_gene_phenotype	hepatocellular_carcinoma
gene_civic_url	.
description	.
---	---
BAGE2	
chrom	chr21
synonym	CT2.2
hgnc_id	15723
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
BCL2	
chrom	chr18
synonym	Bcl-2,PPP1R50
hgnc_id	990
entrez_id	596
rvis_pct	39.67916962
is_OG	1
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/59
description	.
---	---
BCOR	
chrom	chrX
synonym	KIAA1575,FLJ20285
hgnc_id	20893
entrez_id	54880
rvis_pct	0.507195093
is_OG	0
is_TS	0
in_cgi_biomarkers	1

clinvar_gene_phenotype	congenital_cataract| oculofaciocardiodental_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/12555
description	.
---	---
BCR	
chrom	chr22
synonym	BCR1,CML,D22S11,PHL,D22S662,ALL
hgnc_id	1014
entrez_id	613
rvis_pct	5.360934183
is_OG	1
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	chronic_myeloid_leukemia
gene_civic_url	.
description	.
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BIVM-ERCC5	
chrom	chr13
synonym	.
hgnc_id	43690
entrez_id	100533467
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	abnormality_of_the_corpus_callosum| cerebellar_atrophy| cerebrooculofacioskeletal_syndrome_3| cognitive_impairment| dysarthria| pectus_excavatum| pes_cavus| polyneuropathy| spastic_paraplegia| xeroderma_pigmentosum| xeroderma_pigmentosum,group_g| xeroderma_pigmentosum_group_g/cockayne_syndrome
gene_civic_url	.
description	.
---	---
BLM	
chrom	chr15
synonym	RECQ2,RECQL3,BS
hgnc_id	1058
entrez_id	641
rvis_pct	42.36258552
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	bloom_syndrome| hereditary_cancer-predisposing_syndrome

gene_civic_url	.
description	.
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BRAF	
chrom	chr7
synonym	BRAF1
hgnc_id	1097
entrez_id	673
rvis_pct	17.74593064
is_OG	1
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	adenocarcinoma_of_lung| astrocytoma,low-grade,somatic| carcinoma_of_colon| cardio-facio-cutaneous_syndrome| cardiofaciocutaneous_syndrome_1| downslanted_palpebral_fissures| endometrial_carcinoma| germ_cell_tumor,nonseminomatous| high_forehead| leopard_syndrome| leopard_syndrome_3| low-set,posteriorly_rotated_ears| malignant_lymphoma,non-hodgkin| malignant_melanoma| neonatal_respiratory_distress| non-small_cell_lung_cancer| noonan_syndrome| noonan_syndrome_1| noonan_syndrome_7| noonan_syndrome_with_multiple_lentigines| papillary_thyroid_carcinoma| premature_birth| pulmonic_stenosis| rasopathy| thyroid_cancer,follicular| ventricular_hypertrophy| ventricular_septal_defect| webbed_neck| wide_intermamillary_distance
gene_civic_url	https://civic.genome.wustl.edu/links/genes/5
description	BRAF mutations are found to be recurrent in many cancer types. Of these, the mutation of valine 600 to glutamic acid (V600E) is the most prevalent. V600E has been determined to be an activating mutation, and cells that harbor it, along with other V600 mutations are sensitive to the BRAF inhibitor dabrafenib. It is also common to use MEK inhibition as a substitute for BRAF inhibitors, and the MEK inhibitor trametinib has seen some success in BRAF mutant melanomas. BRAF mutations have also been correlated with poor prognosis in many cancer types, although there is at least one study that questions this conclusion in papillary thyroid cancer.
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C11orf30	
chrom	chr11
synonym	EMSY
hgnc_id	18071
entrez_id	56946
rvis_pct	1.875442321
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
CCND1	
chrom	chr11
synonym	D11S287E,BCL1,U21B31,PRAD1
hgnc_id	1582

entrez_id	595
rvis_pct	31.45789101
is_OG	1
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	colorectal_cancer,susceptibility_to| von_hippel-lindau_syndrome,modifier_of
gene_civic_url	https://civic.genome.wustl.edu/links/genes/8
description	Cyclin D has been shown in many cancer types to be misregulated. Well established for their oncogenic properties, the cyclins, and the cyclin-dependent kinases (CDK's) they activate, have been the focus of major research and development efforts over the past decade. The methods by which the cyclins are misregulated are widely variable, ranging from genomic amplification to changes in promoter methylation. While Cyclin D2 has only been found to be significantly deregulated in glioma, Cyclin D1 seems to be a pan-cancer actor. Cyclin D misregulation has been shown to lead to poorer outcomes in a number of studies, but currently there are no FDA-approved targeted therapies.
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CD79A	
chrom	chr19
synonym	MB-1,IGA
hgnc_id	1698
entrez_id	973
rvis_pct	63.00424628
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
CDK8	
chrom	chr13
synonym	K35
hgnc_id	1779
entrez_id	1024
rvis_pct	35.42108988
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
CDKN2A	
chrom	chr9
synonym	p16INK4a,p16,CDKN2,MTS1,INK4,MLM,p14,CMM2,ARF,p19,INK4a,p19Arf,CDK4I

hgnc_id	1787
entrez_id	1029
rvis_pct	85.98136353
is_OG	0
is_TS	1
in_cgi_biomarkers	1
clinvar_gene_phenotype	cutaneous_malignant_melanoma_1| cutaneous_melanoma| hereditary_cancer-predisposing_syndrome| hereditary_cutaneous_melanoma| melanoma-pancreatic_cancer_syndrome| melanoma,cutaneous_malignant,susceptibility_to,2| orolaryngeal_cancer,multiple
gene_civic_url	https://civic.genome.wustl.edu/links/genes/14
description	CDKN2A loss has been shown to be a significant event in a number of cancer types. While no targeted therapeutic has been engaged in clinical trials, the prognostic impact has been studied by a number of meta-analyses. In majority of cases CDKN2A is inactivated by homozygous deletions. One of the mechanisms by which loss of CDKN2A can occur is by hypermethylation of the promoter region for the gene. However, the prognostic impact of promoter hypermethylation has been relatively ambiguous. Many studies have suggesting poorer prognostic outcome for patients with hypermethylation in colorectal, liver, and younger lung cancer patients. This being said, there is still research to be done before this becomes a widely-accepted prognostic indicator. Additionally, CDKN2A (p16) expression is a surrogate marker for HPV infection. As such, CDKN2A expression is prognostic in Oropharyngeal and probably also non-oropharyngeal head and neck squamous cell carcinomas.
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CELF3	
chrom	chr1
synonym	ERDA4,BRUNOL1,TNRC4,CAGH4,MGC57297
hgnc_id	11967
entrez_id	11189
rvis_pct	49.75819769
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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CHD2	
chrom	chr15
synonym	DKFZp686E01200,DKFZp547I1315,FLJ38614,DKFZp781D1727
hgnc_id	1917
entrez_id	1106
rvis_pct	2.37084218
is_OG	0
is_TS	0
in_cgi_biomarkers	.

clinvar_gene_phenotype	abnormal_facial_shape| abnormality_of_the_optic_nerve| abnormality_of_the_pinna| cns_hypomyelination| downturned_corners_of_mouth| epileptic_encephalopathy,childhood-onset| growth_delay| hypoplasia_of_the_corpus_callosum| inborn_genetic_diseases| malignant_tumor_of_prostate| microcephaly| postnatal_microcephaly| sparse_eyebrow| sparse_scalp_hair
gene_civic_url	.
description	.
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CHD4	
chrom	chr12
synonym	Mi-2b,Mi2-BETA
hgnc_id	1919
entrez_id	1108
rvis_pct	2.824958717
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	sifrim-hitz-weiss_syndrome
gene_civic_url	.
description	.
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CHEK2P2	
chrom	chr15
synonym	.
hgnc_id	43578
entrez_id	646096
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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CREBBP	
chrom	chr16
synonym	KAT3A,CBP,RTS,RSTS
hgnc_id	2348
entrez_id	1387
rvis_pct	1.539278132
is_OG	0
is_TS	0
in_cgi_biomarkers	.

clinvar_gene_phenotype	abnormality_of_the_thumb| glaucoma| global_developmental_delay| hirschsprung_disease_1| inborn_genetic_diseases| rubinstein-taybi_syndrome| scoliosis
gene_civic_url	https://civic.genome.wustl.edu/links/genes/1193
description	.
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CSF3R	
chrom	chr1
synonym	CD114,GCSFR
hgnc_id	2439
entrez_id	1441
rvis_pct	96.86836518
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	early_t_cell_progenitor_acute_lymphoblastic_leukemia| hereditary_neutrophilia| neutropenia,severe_congenital,7,autosomal_recessive| severe_congenital_neutropenia
gene_civic_url	https://civic.genome.wustl.edu/links/genes/1239
description	.
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CTNNB1	
chrom	chr3
synonym	CTNNB,beta-catenin,armadillo
hgnc_id	2514
entrez_id	1499
rvis_pct	18.44184949
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	carcinoma_of_colon| desmoid_tumor,somatic| hepatoblastoma| hepatocellular_carcinoma| inborn_genetic_diseases| malignant_tumor_of_prostate| medulloblastoma| mental_retardation,autosomal_dominant_19| neoplasm_of_ovary| pilomatrixoma
gene_civic_url	https://civic.genome.wustl.edu/links/genes/1290
description	.
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CUL3	
chrom	chr2
synonym	.
hgnc_id	2553
entrez_id	8452
rvis_pct	43.29440906
is_OG	0
is_TS	0
in_cgi_biomarkers	.

clinvar_gene_phenotype	pseudohypoaldosteronism,type_i,dominant| pseudohypoaldosteronism,type_2| pseudohypoaldosteronism_type_2e
gene_civic_url	.
description	.
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CUX1	
chrom	chr7
synonym	CUTL1,CUT,GOLIM6,CUX,CDP1,Cux/CDP,CDP,CDP/Cut,CDP/Cux,Clox,CASP
hgnc_id	2557
entrez_id	1523
rvis_pct	2.011087521
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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DCP1B	
chrom	chr12
synonym	FLJ31638
hgnc_id	24451
entrez_id	196513
rvis_pct	85.76315169
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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DENND4B	
chrom	chr1
synonym	KIAA0476
hgnc_id	29044
entrez_id	9909
rvis_pct	0.996697334
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.

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DICER1	
chrom	chr14
synonym	KIAA0928,MNG1,K12H4.8-LIKE,Dicer,HERNA
hgnc_id	17098
entrez_id	23405
rvis_pct	3.44420854
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	anophthalmia_-*microphthalmia| dicer1-related_pleuropulmonary_blastoma_cancer_predisposition_syndrome| goiter,multinodular_1,with_or_without_sertoli-leydig_cell_tumors| pineoblastoma| pleuropulmonary_blastoma| rhabdomyosarcoma,embryonal,2*
gene_civic_url	https://civic.genome.wustl.edu/links/genes/9533
description	.
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DLX6	
chrom	chr7
synonym	.
hgnc_id	2919
entrez_id	1750
rvis_pct	41.24793583
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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DNMT1	
chrom	chr19
synonym	DNMT,CXXC9,MCMT
hgnc_id	2976
entrez_id	1786
rvis_pct	3.326256192
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	acanthocytosis| ataxia| cerebellar_ataxia,deafness,and_narcolepsy,autosomal_dominant| cerebellar_atrophy| cerebral_atrophy| chorea| dementia,deafness,and_sensory_neuropathy| dysarthria| dysphagia| hereditary_sensory_neuropathy_type_ie
gene_civic_url	https://civic.genome.wustl.edu/links/genes/1510
description	.

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DNMT3B	
chrom	chr20
synonym	.
hgnc_id	2979
entrez_id	1789
rvis_pct	2.535975466
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	centromeric_instability_of_chromosomes_1\x2c9_and_16_and_immunodeficiency
gene_civic_url	.
description	.
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DOT1L	
chrom	chr19
synonym	KMT4,DOT1,KIAA1814
hgnc_id	24948
entrez_id	84444
rvis_pct	0.44821892
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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E2F4	
chrom	chr16
synonym	E2F-4
hgnc_id	3118
entrez_id	1874
rvis_pct	13.93606983
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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EP300	
chrom	chr22

synonym	KAT3B,p300
hgnc_id	3373
entrez_id	2033
rvis_pct	0.259495164
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	abnormality_of_the_thumb| carcinoma_of_colon| congenital_microcephaly| facial_grimacing| feeding_difficulties| global_developmental_delay| hirsutism| intellectual_disability,moderate| microcephaly| micrognathia| muscular_hypotonia| myopia| rubinstein-taybi_syndrome| rubinstein-taybi_syndrome_2| short_stature| synophrys| intellectual_deficiency
gene_civic_url	.
description	.
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EP400	
chrom	chr12
synonym	TNRC12,P400,KIAA1498,DKFZP434I225,CAGH32,KIAA1818
hgnc_id	11958
entrez_id	57634
rvis_pct	0.218211842
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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EPHA5	
chrom	chr4
synonym	TYRO4,Hek7,CEK7,EHK1
hgnc_id	3389
entrez_id	2044
rvis_pct	5.490681765
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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EPHA7	
chrom	chr6
synonym	Hek11

hgnc_id	3390
entrez_id	2045
rvis_pct	10.84571833
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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ERBB4	
chrom	chr2
synonym	ALS19
hgnc_id	3432
entrez_id	2066
rvis_pct	1.786978061
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	amyotrophic_lateral_sclerosis_19
gene_civic_url	https://civic.genome.wustl.edu/links/genes/1734
description	ErbB4 (HER4) is one of the four members in the EGFR subfamily of receptor tyrosine kinases. Ligands include EGF, epiregulin, betacellulin and the neuregulins (Sundvall et. al.). Of these, NRG3 and NRG4 exclusively bind HER4 (Hynes et. al.). Mutations in ERBB4 have been identified in various cancer types including melanoma, lung adenocarcinoma and medulloblastoma. A therapeutic value of these aberrations still remains unknown (Arteaga et. al.).
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ERCC2	
chrom	chr19
synonym	MGC126218,MGC102762,MGC126219,XPD,MAG,TFIIH,EM9
hgnc_id	3434
entrez_id	2068
rvis_pct	1.887237556
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	cerebro-oculo-facio-skeletal_syndrome| cerebrooculofacioskeletal_syndrome_2| ercc2-related_disorders| trichothiodystrophy_1,photosensitive| xeroderma_pigmentosum| xeroderma_pigmentosum,group_d
gene_civic_url	https://civic.genome.wustl.edu/links/genes/1736
description	ERCC2 functions as a DNA repair gene involved in separating the double helix via 5’-3’ helicase activity. It forms a part of the transcription factor II Human (TFIIH) complex and is ATP-dependent. The TFIIH complex is known to be involved in the nucleotide excision repair pathway (NER) which can repair DNA damage caused by chemotherapeutic treatment and basal transcription. ERCC2 variants have been observed in a variety of cancers. A number of studies have suggested ERCC2 variants can act as biomarkers to predict response to neoadjuvant treatment, and cancer prognosis. Additionally the Lys751Gln polymorphism has been observed to increase risk in a number of cancer types; however, results have been conflicting.

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ESR1	
chrom	chr6
synonym	NR3A1,ESR,Era
hgnc_id	3467
entrez_id	2099
rvis_pct	69.45623968
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	cerebellar_ataxia| emery-dreifuss_muscular_dystrophy| estrogen_receptor_mutant,temperature-sensitive| estrogen_resistance| familial_cancer_of_breast
gene_civic_url	https://civic.genome.wustl.edu/links/genes/21
description	ESR1 has been a focus in breast cancer for quite some time, but is also clinically relevant in endometrial, ovarian and other cancer types. The identification of ER-positive breast cancers that are resistant to hormone therapy have inspired clinical sequencing efforts to shed light on the mechanisms of this resistance. A number of mutations in the ligand binding domain of ESR1 have been implicated in hormone resistance and anti-estrogen therapies. These observations have spurred efforts to develop therapeutics that stimulate ESR1 protein degradation (e.g. Fulvestrant), rather than acting as a small molecule antagonist. These agents are currently in clinical trials and have seen some success.
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ETV5	
chrom	chr3
synonym	ERM
hgnc_id	3494
entrez_id	2119
rvis_pct	29.16371786
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/1768
description	.
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FANCD2	
chrom	chr3
synonym	FANCD,FAD,FACD,FA-D2
hgnc_id	3585
entrez_id	2177
rvis_pct	9.790044822
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	fanconi_anemia| fanconi_anemia,complementation_group_d2

gene_civic_url	.
description	.
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FANCE	
chrom	chr6
synonym	FACE,FAE
hgnc_id	3586
entrez_id	2178
rvis_pct	32.05944798
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	fanconi_anemia| fanconi_anemia,complementation_group_e
gene_civic_url	.
description	.
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FAS	
chrom	chr10
synonym	APO-1,FAS1,CD95,APT1,TNFRSF6
hgnc_id	11920
entrez_id	355
rvis_pct	78.46190139
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	autoimmune_lymphoproliferative_syndrome| autoimmune_lymphoproliferative_syndrome,type_1a| lung_cancer,susceptibility_to| moyamoya_disease| multisystemic_smooth_muscle_dysfunction_syndrome| squamous_cell_carcinoma,burn_scar-related,somatic| thoracic_aortic_aneurysm_and_aortic_dissection
gene_civic_url	.
description	.
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FBXW7	
chrom	chr4
synonym	AGO,FBW7,SEL-10,FBX30,SEL10,FLJ11071,CDC4,FBXW6
hgnc_id	16712
entrez_id	55294
rvis_pct	45.35857514
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/12903

description	.
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FLCN	
chrom	chr17
synonym	MGC23445,BHD,MGC17998
hgnc_id	27310
entrez_id	201163
rvis_pct	39.17197452
is_OG	0
is_TS	1
in_cgi_biomarkers	1
clinvar_gene_phenotype	birt-hogg-dub_syndrome| carcinoma_of_colon| hereditary_cancer-predisposing_syndrome| multiple_fibrofolliculomas| pneumothorax,primary_spontaneous| spontaneous_pneumothorax
gene_civic_url	https://civic.genome.wustl.edu/links/genes/19959
description	.
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FLT1	
chrom	chr13
synonym	VEGFR1,FLT
hgnc_id	3763
entrez_id	2321
rvis_pct	3.44420854
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	carcinoma_of_colon
gene_civic_url	.
description	.
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FLT4	
chrom	chr5
synonym	VEGFR3,PCL
hgnc_id	3767
entrez_id	2324
rvis_pct	85.53314461
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	carcinoma_of_colon| hemangioma,capillary_infantile| hereditary_lymphedema_type_i
gene_civic_url	https://civic.genome.wustl.edu/links/genes/1938
description	.
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FOXP1	
chrom	chr3
synonym	12CC4,QRF1,hFKH1B,HSPC215
hgnc_id	3823
entrez_id	27086
rvis_pct	28.01368247
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	anterior_creases_of_earlobe| atrioventricular_septal_defect| autism| congenital_atresia_of_aortic_valve| congenital_atresia_of_mitral_valve| delayed_speech_and_language_development| glabellar_hemangioma| heterotaxy_syndrome| hypoplastic_left_heart_syndrome| intellectual_disability_with_language_impairment_and_autistic_features| intellectual_disability| mental_retardation_with_language_impairment_and_with_or_without_autistic_features| pulmonary_atresia_with_ventricular_septal_defect| single_ventricle_defect| strabismus
gene_civic_url	https://civic.genome.wustl.edu/links/genes/56
description	.
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FOXP2	
chrom	chr7
synonym	TNRC10,CAGH44,SPCH1
hgnc_id	13875
entrez_id	93986
rvis_pct	11.67728238
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	speech-language_disorder_1
gene_civic_url	.
description	.
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GATA1	
chrom	chrX
synonym	ERYF1,GF1,GATA-1,NF-E1,NFE1
hgnc_id	4170
entrez_id	2623
rvis_pct	17.74593064
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	acute_megakaryoblastic_leukemia| diamond-blackfan_anemia| dyserythropoietic_anemia_with_thrombocytopenia| gata-1-related_thrombocytopenia_with_dyserythropoiesis| leukemia,megakaryoblastic,of_down_syndrome| thrombocytopenia,x-linked,without_dyserythropoietic_anemia| thrombocytopenia,platelet_dysfunction,hemolysis,and_imbalanced_globin_synthesis

gene_civic_url	.
description	.
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GATA2	
chrom	chr3
synonym	NFE1B
hgnc_id	4171
entrez_id	2624
rvis_pct	25.56027365
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	dendritic_cell,monocyte,b_lymphocyte,and_natural_killer_lymphocyte_deficiency| leukemia,acute_myeloid,susceptibility_to| lymphedema,primary,with_myelodysplasia| myelodysplastic_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/25
description	GATA2 is a transcription factor involved in stem cell maintenance with key roles in hematopoietic development. GATA2 mutations are associated with a variety of inherited and acquired immune disorders including myelodysplastic syndrome and acute myeloid leukemia. In addition to a role in hematopoiesis, the maintenance GATA2 expression has been implicated as a requirement in KRAS-driven non-small cell lung cancer. Preclinical models have indicated therapeutic benefit from targeting GATA2-mediated pathways in the context of KRAS-driven NSCLC.
---	---
GLI1	
chrom	chr12
synonym	GLI
hgnc_id	4317
entrez_id	2735
rvis_pct	10.5626327
is_OG	1
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	malignant_tumor_of_prostate
gene_civic_url	https://civic.genome.wustl.edu/links/genes/2279
description	.
---	---
GNAQ	
chrom	chr9
synonym	G-ALPHA-q,GAQ
hgnc_id	4390
entrez_id	2776
rvis_pct	35.42108988
is_OG	0
is_TS	0
in_cgi_biomarkers	1

clinvar_gene_phenotype	capillary_malformations,congenital| sturge-weber_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/2317
description	.
---	---
GNAS	
chrom	chr20
synonym	SCG6,NESP,GPSA,NESP55,GNASXL,GNAS1
hgnc_id	4392
entrez_id	2778
rvis_pct	52.31776362
is_OG	1
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	brachydactyly_syndrome| cognitive_impairment| cushing's_syndrome| hypocalcemia| inborn_genetic_diseases| mccune-albright_syndrome| obesity| pseudohypoparathyroidism,type_ia,with_testotoxicosis| pituitary_dependent_hypercortisolism| polyostotic_fibrous_dysplasia,somatic,mosaic| progressive_osseous_heteroplasia| pseudohypoparathyroidism_type_1a| pseudohypoparathyroidism_type_1c| pseudopseudohypoparathyroidism| round_face| sex_cord-stromal_tumor| short_stature| somatotroph_adenoma| subcutaneous_nodule| tetany
gene_civic_url	https://civic.genome.wustl.edu/links/genes/2319
description	.
---	---
HIST1H1E	
chrom	chr6
synonym	H1F4,H1e,H1s-4,H1.4
hgnc_id	4718
entrez_id	3008
rvis_pct	5.832743572
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
HIST1H3E	
chrom	chr6
synonym	H3.1,H3/d,H3FD
hgnc_id	4769
entrez_id	8350
rvis_pct	17.74593064
is_OG	0
is_TS	0
in_cgi_biomarkers	.

clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
HIST2H2BE	
chrom	chr1
synonym	H2BFQ,H2B,H2B.1,H2B/q
hgnc_id	4760
entrez_id	8349
rvis_pct	64.11299835
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
HIST3H3	
chrom	chr1
synonym	H3/g,H3FT,H3t,H3.4
hgnc_id	4778
entrez_id	8290
rvis_pct	21.41424864
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
HNF1A	
chrom	chr12
synonym	MODY3,TCF1,HNF1,LFB1
hgnc_id	11621
entrez_id	6927
rvis_pct	24.59896202
is_OG	0
is_TS	0
in_cgi_biomarkers	.

clinvar_gene_phenotype	chromophobe_renal_cell_carcinoma| clear_cell_carcinoma_of_kidney| diabetes_mellitus,type_ii,susceptibility_to| diabetes_mellitus,insulin-dependent,20| diabetes_mellitus_type_1| diabetes_mellitus_type_2| hepatic_adenomas,familial| insulin_resistance,susceptibility_to| maturity-onset_diabetes_of_the_young| maturity-onset_diabetes_of_the_young,type_3| maturity-onset_diabetes_of_the_young,type_3,maturity-onset_diabetes_of_the_young,type_3| serum_hdl_cholesterol_level,modifier_of
gene_civic_url	.
description	.
---	---
HSP90AA1	
chrom	chr14
synonym	HSP90N,HSPC1,Hsp90,HSPCA,FLJ31884,Hsp89
hgnc_id	5253
entrez_id	3320
rvis_pct	8.144609578
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
HTT	
chrom	chr4
synonym	IT15,HD
hgnc_id	4851
entrez_id	3064
rvis_pct	1.692616183
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
IGF1R	
chrom	chr15
synonym	IGFR,IGFIR,CD221,JTK13,MGC18216
hgnc_id	5465
entrez_id	3480
rvis_pct	2.494692144
is_OG	0
is_TS	0
in_cgi_biomarkers	1

clinvar_gene_phenotype	insulin-like_growth_factor_i_resistance| insulin-like_growth_factor_1_resistance_to
gene_civic_url	https://civic.genome.wustl.edu/links/genes/2899
description	.
---	---
INHA	
chrom	chr2
synonym	.
hgnc_id	6065
entrez_id	3623
rvis_pct	60.31493277
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	three_m_syndrome
gene_civic_url	.
description	.
---	---
KCNN3	
chrom	chr1
synonym	SKCA3,KCa2.3,hSK3
hgnc_id	6292
entrez_id	3782
rvis_pct	19.54470394
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	malignant_tumor_of_prostate
gene_civic_url	.
description	.
---	---
KDM5A	
chrom	chr12
synonym	RBBP2,JARID1A
hgnc_id	9886
entrez_id	5927
rvis_pct	13.78862939
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.

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KDM5C	
chrom	chrX
synonym	XE169,SMCX,MRX13,DXS1272E,JARID1C
hgnc_id	11114
entrez_id	8242
rvis_pct	3.503184713
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	intellectual_disability| mental_retardation,syndromic,claes-jensen_type,x-linked| spastic_paraplegia
gene_civic_url	https://civic.genome.wustl.edu/links/genes/6538
description	.
---	---
KDM6A	
chrom	chrX
synonym	UTX
hgnc_id	12637
entrez_id	7403
rvis_pct	48.77919321
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	kabuki_syndrome_2| malignant_tumor_of_prostate
gene_civic_url	.
description	.
---	---
KEL	
chrom	chr7
synonym	CD238,ECE3
hgnc_id	6308
entrez_id	3792
rvis_pct	3.998584572
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	kell_k/k_blood_group_polymorphism| kel6_antigen
gene_civic_url	.
description	.
---	---
KIAA1462	
chrom	chr10

synonym	JCAD
hgnc_id	29283
entrez_id	57608
rvis_pct	93.16466148
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
KIAA2018	
chrom	chr3
synonym	.
hgnc_id	30494
entrez_id	205717
rvis_pct	14.50224109
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
KMT2B	
chrom	chr19
synonym	TRX2,HRX2,MLL1B,KIAA0304,MLL2,MLL4,WBP7
hgnc_id	15840
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
KMT2C	
chrom	chr7
synonym	MLL3,HALR,KIAA1506
hgnc_id	13726
entrez_id	58508

rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/14089
description	.
---	---
LAMP1	
chrom	chr13
synonym	CD107a
hgnc_id	6499
entrez_id	3916
rvis_pct	39.11299835
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
LMO1	
chrom	chr11
synonym	TTG1,RHOM1,RBTN1
hgnc_id	6641
entrez_id	4004
rvis_pct	41.24793583
is_OG	1
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	lim_domain_only-1_polymorphism
gene_civic_url	.
description	.
---	---
LRP1B	
chrom	chr2
synonym	LRP-DIT,LRPDIT
hgnc_id	6693
entrez_id	53353
rvis_pct	1.226704411
is_OG	0
is_TS	0

in_cgi_biomarkers	1
clinvar_gene_phenotype	malignant_tumor_of_prostate
gene_civic_url	https://civic.genome.wustl.edu/links/genes/12146
description	LRP1B is a putative tumor suppressor and a member of the low-density lipoprotein (LDL) receptor family. The LDL receptor family have roles related to clearance of extracellular ligand and are proposed to be involved in extracellular signal transduction. silencing and down-expression of LRP1B as been observed in renal cell carcinoma and thyroid cancer. Further Deletion of LRP1B has been associated with chemotherapy resistance in high-grade serous cancers.
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LYN	
chrom	chr8
synonym	JTK8
hgnc_id	6735
entrez_id	4067
rvis_pct	14.96815287
is_OG	1
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/3359
description	.
---	---
MAGI1	
chrom	chr3
synonym	WWP3,BAIAP1,TNRC19,AIP3,MAGI-1,BAP1
hgnc_id	946
entrez_id	9223
rvis_pct	4.358339231
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
MAP3K1	
chrom	chr5
synonym	MEKK1,MEKK,MAPKKK1
hgnc_id	6848
entrez_id	4214
rvis_pct	5.520169851
is_OG	0
is_TS	0
in_cgi_biomarkers	.

clinvar_gene_phenotype	46\x2cxy_sex_reversal,type_6| anophthalmia-*microphthalmia*
gene_civic_url	.
description	.
---	---
MAP3K4	
chrom	chr6
synonym	KIAA0213,MTK1,MEKK4,MAPKKK4
hgnc_id	6856
entrez_id	4216
rvis_pct	7.324840764
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
MED12	
chrom	chrX
synonym	KIAA0192,TNRC11,TRAP230,OKS,FGS1,CAGH45,OPA1,HOPA
hgnc_id	11957
entrez_id	9968
rvis_pct	8.47487615
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	abnormal_facial_shape| absent_speech| agenesis_of_corpus_callosum| anal_atresia| broad_thumb| cardiovascular_phenotype| expressive_language_delay| fg_syndrome| global_developmental_delay| inborn_genetic_diseases| intellectual_disability| microcephaly| ohdo_syndrome,x-linked| seizures| uterine_leiomyoma| ventriculomegaly| x-linked_mental_retardation_with_marfanoid_habitus_syndrome
gene_civic_url	.
description	.
---	---
MLH1	
chrom	chr3
synonym	HNPCC2,COCA2,FCC2,HNPCC
hgnc_id	7127
entrez_id	4292
rvis_pct	80.29606039
is_OG	0
is_TS	1
in_cgi_biomarkers	.

clinvar_gene_phenotype	colorectal_cancer,non-polyposis| colorectal_cancer,sporadic,susceptibility_to| endometrial_carcinoma| hereditary_cancer-predisposing_syndrome| lynch_syndrome| lynch_syndrome_i| lynch_syndrome_ii| ovarian_cancer| turcot_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/3532
description	MLH1 is a tumor suppressor gene involved in DNA mismatch repair. Germline mutations in this gene are known to cause Lynch syndrome. The most common malignancies in Lynch syndrome are colorectal and endometrial carcinomas. In addition to germline mutations, somatic mutations in this gene have been described in colorectal and endometrial cancers.
---	---
MLLT3	
chrom	chr9
synonym	YEATS3,AF-9,AF9
hgnc_id	7136
entrez_id	4300
rvis_pct	28.93371078
is_OG	1
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
MN1	
chrom	chr22
synonym	MGCR,MGCR1,MGCR1-PEN
hgnc_id	7180
entrez_id	4330
rvis_pct	.
is_OG	0
is_TS	1
in_cgi_biomarkers	.
clinvar_gene_phenotype	inborn_genetic_diseases
gene_civic_url	.
description	.
---	---
MRE11A	
chrom	chr11
synonym	MRE11,ATLD
hgnc_id	7230
entrez_id	4361
rvis_pct	57.40740741
is_OG	0
is_TS	0
in_cgi_biomarkers	.

clinvar_gene_phenotype	ataxia-telangiectasia-like_disorder_1| hereditary_cancer-predisposing_syndrome| malignant_tumor_of_urinary_bladder| ovarian_neoplasm| triple-negative_breast_cancer| tumor_susceptibility_linked_to_germline_bap1_mutations
gene_civic_url	.
description	.
---	---
MSH2	
chrom	chr2
synonym	HNPCC,COCA1,HNPCC1
hgnc_id	7325
entrez_id	4436
rvis_pct	1.680820948
is_OG	0
is_TS	1
in_cgi_biomarkers	.
clinvar_gene_phenotype	colorectal_cancer| colorectal_cancer,hereditary,nonpolyposis,type_1| colorectal_cancer,non-polyposis| hereditary_cancer-predisposing_syndrome| lynch_syndrome| lynch_syndrome_i| msh2_polymorphism| muir-torr_syndrome| ovarian_cancer| renal_cell_carcinoma| tumor_susceptibility_linked_to_germline_bap1_mutations| turcot_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/3628
description	.
---	---
MSH3	
chrom	chr5
synonym	DUP,MRP1
hgnc_id	7326
entrez_id	4437
rvis_pct	47.25760793
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	endometrial_carcinoma| familial_adenomatous_polyposis_4| gastrointestinal_stromal_tumor
gene_civic_url	.
description	.
---	---
MSH6	
chrom	chr2
synonym	GTBP
hgnc_id	7329
entrez_id	2956
rvis_pct	1.279782968
is_OG	0
is_TS	0
in_cgi_biomarkers	.

clinvar_gene_phenotype	colorectal/*endometrial_cancer| colorectal_cancer| colorectal_cancer,early_onset| colorectal_cancer,non-polyposis| endometrial_carcinoma| hereditary_cancer-predisposing_syndrome| hereditary_nonpolyposis_colorectal_cancer_type_5| hereditary_nonpolyposis_colorectal_carcinoma| lynch_syndrome| lynch_syndrome_i| ovarian_cancer| tumor_susceptibility_linked_to_germline_bap1_mutations| turcot_syndrome*
gene_civic_url	https://civic.genome.wustl.edu/links/genes/2478
description	.
---	---
MST1	
chrom	chr3
synonym	D3F15S2,NF15S2,HGFL,DNF15S2,MSP
hgnc_id	7380
entrez_id	4485
rvis_pct	25.79028073
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
MST1P2	
chrom	chr1
synonym	MSPL-2,MSTP2,MSPL2
hgnc_id	7383
entrez_id	101930052
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
MST1R	
chrom	chr3
synonym	CDw136,PTK8,RON,CD136
hgnc_id	7381
entrez_id	4486
rvis_pct	2.288275537
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	nasopharyngeal_carcinoma,susceptibility_to,3

gene_civic_url	.
description	.
---	---
MTOR	
chrom	chr1
synonym	RAFT1,FRAP1,FRAP2,FLJ44809,RAPT1,FRAP
hgnc_id	3942
entrez_id	2475
rvis_pct	0.342061807
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	smith-kingsmore_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/2073
description	mTOR deregulation has been observed in many cancer types. As a part of the PI3K/ Akt pathway, there is a broad interest in mTOR biology across cancer types. mTOR inhibition has been investigated for nearly a decade. Everolimus, temsirolimus and zotarolimus are three of the more commonly used mTOR inhibitors used in clinical treatment today, with modest success.
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MYCN	
chrom	chr2
synonym	MYCNOT,NMYC,N-myc,bHLHe37
hgnc_id	7559
entrez_id	4613
rvis_pct	27.41802312
is_OG	1
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	feingold_syndrome_1
gene_civic_url	https://civic.genome.wustl.edu/links/genes/3741
description	.
---	---
NCOA3	
chrom	chr20
synonym	AIB1,TNRC16,RAC3,p/CIP,ACTR,TRAM-1,SRC3,SRC-3,KAT13B,CAGH16,bHLHe42
hgnc_id	7670
entrez_id	8202
rvis_pct	5.537862703
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/74

description	.
---	---
NCOA6	
chrom	chr20
synonym	RAP250,KIAA0181,AIB3,NRC,TRBP,ASC2,PRIP
hgnc_id	15936
entrez_id	23054
rvis_pct	1.710309035
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
NCOR1	
chrom	chr17
synonym	MGC104216,hCIT529I10,N-CoR,TRAC1,KIAA1047,hN-CoR
hgnc_id	7672
entrez_id	9611
rvis_pct	1.49799481
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	bilateral_cleft_lip_and_palate| camptodactyly_of_finger| familial_cancer_of_breast| hypertelorism| low-set_ears| postaxial_hand_polydactyly| premature_birth| scrotal_hypoplasia| wide_intermamillary_distance| zunich_neuroectodermal_syndrome
gene_civic_url	.
description	.
---	---
NCOR1P2	
chrom	chr17
synonym	.
hgnc_id	42997
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---

NCOR2

chrom	chr12
synonym	SMRT,SMRTE,TRAC-1,CTG26,TNRC14
hgnc_id	7673
entrez_id	9612
rvis_pct	0.819768813
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---

NF1P3

chrom	chr21
synonym	NF1L1
hgnc_id	7766
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---

NF1P6

chrom	chr22
synonym	NF1L6,Em:AP000532.C22.1
hgnc_id	7771
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---

NKX3-1

chrom	chr8
synonym	NKX3A,BAPX2,NKX3.1

hgnc_id	7838
entrez_id	4824
rvis_pct	.
is_OG	0
is_TS	1
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
NOTCH1	
chrom	chr9
synonym	TAN1
hgnc_id	7881
entrez_id	4851
rvis_pct	0.330266572
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	adams-oliver_syndrome_1| adams-oliver_syndrome_5| anophthalmia-*microphthalmia| aortic_valve_disorder| arterial_dissection| cardiovascular_phenotype| early_t_cell_progenitor_acute_lymphoblastic_leukemia| ehlers-danlos_syndrome,type_3| heart,malformation_of| marfan_syndrome| tetralogy_of_fallot| thoracic_aortic_aneurysm_and_aortic_dissection*
gene_civic_url	https://civic.genome.wustl.edu/links/genes/50
description	NOTCH1 is one of four known genes encoding the NOTCH family of proteins, a group of receptors involved in the Notch signaling pathway. NOTCH proteins are characterized by N-terminal EGF-like repeats followed by LNR domains which form a complex with ligands to prevent signaling. The Notch signaling pathway is involved in processes related to cell fate specification, differentiation, proliferation, and survival. Activation of Notch has been shown to be correlative with mammary tumorgenesis in mice and increased expression of Notch receptors has been observed in a variety of cancer types including cervical, colon, head and neck, lung, renal, pancreatic, leukemia, and breast cancer. A number of treatment modalities have been explored related to Notch inhibition especially in breast cancer with mixed results.
---	---
NOTCH2	
chrom	chr1
synonym	.
hgnc_id	7882
entrez_id	4853
rvis_pct	2.14673272
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	alagille_syndrome_2| hajdu-cheney_syndrome| monoclonal_b-cell_lymphocytosis
gene_civic_url	.
description	.

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NPM1	
chrom	chr5
synonym	NPM,B23
hgnc_id	7910
entrez_id	4869
rvis_pct	30.06605331
is_OG	1
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	acute_myeloid_leukemia| myelodysplastic_syndrome_progressed_to_acute_myeloid_leukemia
gene_civic_url	https://civic.genome.wustl.edu/links/genes/35
description	AML with mutated NPM1 is a provisional entity in the WHO classification of AML and is recommended to be tested in patients with cytogenetically normal AML (CN-AML). FLT3 mutations should be evaluated concurrently as they have prognostic consequences. NPM1 mutations are concentrated in exon 12, most frequently W288fs which results in cytoplasmic sequestration of the protein. Exon 12 NPM1 mutations in the absence of FLT3-ITD are associated with good prognostic outcomes. Mice expressing the Npm1-W288fs mutation develop myeloproliferative neoplasms but not overt leukemia, indicating it may require additional mutations to promote leukemic development.
---	---
NTRK1	
chrom	chr1
synonym	MTC,TRK,TRKA
hgnc_id	8031
entrez_id	4914
rvis_pct	48.90894079
is_OG	1
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	familial_medullary_thyroid_carcinoma| hereditary_insensitivity_to_pain_with_anhidrosis| inborn_genetic_diseases
gene_civic_url	https://civic.genome.wustl.edu/links/genes/3983
description	.
---	---
NTRK2	
chrom	chr9
synonym	TRKB
hgnc_id	8032
entrez_id	4915
rvis_pct	22.64685067
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	obesity,hyperphagia,and_developmental_delay

gene_civic_url	https://civic.genome.wustl.edu/links/genes/3984
description	.
---	---
NUP93	
chrom	chr16
synonym	KIAA0095
hgnc_id	28958
entrez_id	9688
rvis_pct	4.134229771
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	nephrotic_syndrome,type_12
gene_civic_url	.
description	.
---	---
PARP1	
chrom	chr1
synonym	PPOL,ADPRT,PARP
hgnc_id	270
entrez_id	142
rvis_pct	8.982071243
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/199
description	.
---	---
PAX5	
chrom	chr9
synonym	BSAP
hgnc_id	8619
entrez_id	5079
rvis_pct	53.72729417
is_OG	1
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	leukemia,acute_lymphoblastic,susceptibility_to,3
gene_civic_url	.
description	.
---	---

PBRM1

chrom	chr3
synonym	PB1,BAF180
hgnc_id	30064
entrez_id	55193
rvis_pct	1.480301958
is_OG	0
is_TS	1
in_cgi_biomarkers	1
clinvar_gene_phenotype	clear_cell_carcinoma_of_kidney
gene_civic_url	https://civic.genome.wustl.edu/links/genes/62
description	.
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PGR

chrom	chr11
synonym	PR,NR3C3
hgnc_id	8910
entrez_id	5241
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/76
description	.
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PIK3CD

chrom	chr1
synonym	p110D
hgnc_id	8977
entrez_id	5293
rvis_pct	2.724699222
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	immunodeficiency_14
gene_civic_url	.
description	.
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PIK3R1

chrom	chr5
synonym	GRB1,p85-ALPHA,p85

hgnc_id	8979
entrez_id	5295
rvis_pct	13.44656759
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	agammaglobulinemia_7,autosomal_recessive| immunodeficiency_36| malignant_tumor_of_prostate| short_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/4289
description	.
---	---
PLCG2	
chrom	chr16
synonym	.
hgnc_id	9066
entrez_id	5336
rvis_pct	17.91696155
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	autoinflammation,antibody_deficiency,and_immune_dysregulation,plcg2-associated
gene_civic_url	https://civic.genome.wustl.edu/links/genes/4327
description	.
---	---
PMS2	
chrom	chr7
synonym	H_DJ0042M02.9,HNPCC4,PMSL2
hgnc_id	9122
entrez_id	5395
rvis_pct	95.28780373
is_OG	0
is_TS	1
in_cgi_biomarkers	.
clinvar_gene_phenotype	colorectal_cancer,non-polyposis| hereditary_cancer-predisposing_syndrome| hereditary_nonpolyposis_colorectal_cancer_type_4| lynch_syndrome| lynch_syndrome_i| tumor_susceptibility_linked_to_germline_bap1_mutations| turcot_syndrome| not_specified,not_specified
gene_civic_url	https://civic.genome.wustl.edu/links/genes/4371
description	.
---	---
POLD1	
chrom	chr19
synonym	CDC2,POLD
hgnc_id	9175

entrez_id	5424
rvis_pct	11.54163718
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	carcinoma_of_colon| colorectal_cancer| colorectal_cancer_10| hereditary_cancer-predisposing_syndrome| mandibular_hypoplasia,deafness,progeroid_features,and_lipodystrophy_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/4384
description	.
---	---
POLE	
chrom	chr12
synonym	POLE1
hgnc_id	9177
entrez_id	5426
rvis_pct	12.09011559
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	colorectal_cancer| colorectal_cancer,susceptibility_to,12| facial_dysmorphism,immunodeficiency,livedo,and_short_stature| hereditary_cancer-predisposing_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/4386
description	.
---	---
PTCH1	
chrom	chr9
synonym	NBCCS,PTCH,BCNS
hgnc_id	9585
entrez_id	5727
rvis_pct	0.707714084
is_OG	0
is_TS	1
in_cgi_biomarkers	1
clinvar_gene_phenotype	anophthalmia-_microphthalmia| basal_cell_carcinoma,somatic| congenital_heart_disease| gorlin_syndrome| hereditary_cancer-predisposing_syndrome| hirschsprung_disease_1| holoprosencephaly| holoprosencephaly_7| holoprosencephaly_sequence| inborn_genetic_diseases| peters_anomaly| rieger_anomaly
gene_civic_url	https://civic.genome.wustl.edu/links/genes/4645
description	.
---	---
PTEN	
chrom	chr10
synonym	BZS,PTEN1,TEP1,MHAM,MMAC1

hgnc_id	9588
entrez_id	5728
rvis_pct	36.86010852
is_OG	0
is_TS	1
in_cgi_biomarkers	1
clinvar_gene_phenotype	acute_megakaryoblastic_leukemia| bannayan-riley-ruvalcaba_syndrome| cowden_syndrome| cowden_syndrome_1| endometrial_carcinoma| glioma_susceptibility_2| hereditary_cancer-predisposing_syndrome| inborn_genetic_diseases| lhermitte-duclos_disease| macrocephaly/autism_syndrome| malignant_melanoma| mediastinal_germ_cell_tumor| meningioma| pten_hamartoma_tumor_syndrome| prostate_cancer,somatic| proteus-like_syndrome| squamous_cell_carcinoma_of_the_head_and_neck| tumor_susceptibility_linked_to_germline_bap1_mutations| vater_association_with_macrocephaly_and_ventriculomegaly
gene_civic_url	https://civic.genome.wustl.edu/links/genes/41
description	PTEN is a multi-functional tumor suppressor that is very commonly lost in human cancer. Observed in prostate cancer, glioblastoma, endometrial, lung and breast cancer to varying degrees. Up to 70% of prostate cancer patients have been observed to have loss of expression of the gene. It is a part of the PI3K/AKT/mTOR pathway and mTOR inhibitors have been relatively ineffective in treating patients with PTEN loss. New appoaches using microRNAs are currently being investigated.
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PTPRD	
chrom	chr9
synonym	HPTP,PTPD
hgnc_id	9668
entrez_id	5789
rvis_pct	0.483604624
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	malignant_tumor_of_prostate
gene_civic_url	https://civic.genome.wustl.edu/links/genes/4692
description	.
---	---
PTPRS	
chrom	chr19
synonym	.
hgnc_id	9681
entrez_id	5802
rvis_pct	0.100259495
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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QKI	
chrom	chr6
synonym	QK3
hgnc_id	21100
entrez_id	9444
rvis_pct	36.86010852
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
RAD21	
chrom	chr8
synonym	hHR21,SCC1,KIAA0078
hgnc_id	9811
entrez_id	5885
rvis_pct	21.41424864
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	cornelia_de_lange_syndrome_4
gene_civic_url	.
description	.
---	---
RAD50	
chrom	chr5
synonym	hRad50,RAD50-2
hgnc_id	9816
entrez_id	10111
rvis_pct	21.83887709
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	familial_cancer_of_breast| hereditary_cancer-predisposing_syndrome| malignant_tumor_of_prostate| nijmegen_breakage_syndrome-like_disorder
gene_civic_url	https://civic.genome.wustl.edu/links/genes/8032
description	.
---	---
RAD51B	
chrom	chr14
synonym	REC2,RAD51L1,hREC2,R51H2

hgnc_id	9822
entrez_id	5890
rvis_pct	89.79122435
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	hereditary_cancer-predisposing_syndrome
gene_civic_url	.
description	.
---	---
RAD54L	
chrom	chr1
synonym	hHR54,hRAD54,RAD54A
hgnc_id	9826
entrez_id	8438
rvis_pct	28.20240623
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	colonic_adenocarcinoma| ductal_breast_carcinoma| inborn_genetic_diseases| malignant_lymphoma,non-hodgkin
gene_civic_url	.
description	.
---	---
RAI1	
chrom	chr17
synonym	DKFZP434A139,SMCR,MGC12824,SMS,KIAA1820
hgnc_id	9834
entrez_id	10743
rvis_pct	0.253597547
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	deafness,autosomal_recessive_9| intellectual_disability| smith-magenis_syndrome
gene_civic_url	.
description	.
---	---
RANBP2	
chrom	chr2
synonym	ANE1,NUP358,ADANE
hgnc_id	9848
entrez_id	5903
rvis_pct	3.33215381

is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	encephalopathy,acute,infection-induced,3,suceptibility_to
gene_civic_url	.
description	.
---	---
RARG	
chrom	chr12
synonym	RARC,NR1B3
hgnc_id	9866
entrez_id	5916
rvis_pct	34.31823543
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	peters_anomaly
gene_civic_url	.
description	.
---	---
RBM10	
chrom	chrX
synonym	ZRANB5,GPATC9,KIAA0122,DXS8237E,GPATCH9
hgnc_id	9896
entrez_id	8241
rvis_pct	15.76433121
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	tarp_syndrome
gene_civic_url	.
description	.
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RET	
chrom	chr10
synonym	RET51,CDHF12,HSCR1,PTC,CDHR16,MEN2B,MEN2A,MTC1
hgnc_id	9967
entrez_id	5979
rvis_pct	3.001887238
is_OG	1
is_TS	0
in_cgi_biomarkers	1

clinvar_gene_phenotype	central_hypoventilation_syndrome,congenital,with_hirschsprung_disease| congenital_central_hypoventilation| constipation| elevated_basal_serum_calcitonin| fmtc_and_unclassified| familial_medullary_thyroid_carcinoma| gingival_overgrowth| hereditary_cancer-predisposing_syndrome| hirschsprung_disease,dominant| hirschsprung_disease| hirschsprung_disease,protection_against| hirschsprung_disease_1| hypertelorism| hypothyroidism| joint_hypermobility| men2a_and_fmtc| men2a_and_unclassified| men2_phenotype:_unclassified| men2_phenotype:_unknown| multiple_endocrine_neoplasia,type_iia,with_hirschsprung_disease| medullary_thyroid_carcinoma| multiple_endocrine_neoplasia| multiple_endocrine_neoplasia,type_2| multiple_endocrine_neoplasia,type_2,not_specified| multiple_endocrine_neoplasia,type_2a| multiple_endocrine_neoplasia,type_2b| multiple_endocrine_neoplasia_iia| no_men2_disease| pheochromocytoma| renal_adysplasia| short_stature| tetralogy_of_fallot| thick_vermilion_border| thyroid_carcinoma,sporadic_medullary| unclassifed| not_specified,not_specified
gene_civic_url	https://civic.genome.wustl.edu/links/genes/42
description	RET mutations and the RET fusion RET-PTC lead to activation of this tyrosine kinase receptor and are associated with thyroid cancers. RET point mutations are the most common mutations identified in medullary thyroid cancer (MTC) with germline and somatic mutations in RET associated with hereditary and sporadic forms, respectively. The most common somatic form mutation is M918T (exon 16) and a variety of other mutations effecting exons 10, 11 and 15 have been described. The prognostic significance of these mutations have been hotly debated in the field, however, data suggests that some RET mutation may confer drug resistence. No RET-specific agents are currently clinically available but several promiscuous kinase inhibitors that target RET, among others, have been approved for MTC treatment.
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RFWD2	
chrom	chr1
synonym	COP1,FLJ10416,RNF200
hgnc_id	17440
entrez_id	64326
rvis_pct	38.57631517
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
RGPD3	
chrom	chr2
synonym	RGP3
hgnc_id	32416
entrez_id	653489
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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RGPD4

chrom	chr2
synonym	RGP4,DKFZp686P0288
hgnc_id	32417
entrez_id	285190
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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RGPD8

chrom	chr2
synonym	RANBP2L1,RanBP2alpha
hgnc_id	9849
entrez_id	84220
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
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RICTOR

chrom	chr5
synonym	PIA,MGC39830,KIAA1999,AVO3
hgnc_id	28611
entrez_id	253260
rvis_pct	33.55744279
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/20480
description	.
---	---

RNF43

chrom	chr17
synonym	DKFZp781H0392,FLJ20315,URCC

hgnc_id	18505
entrez_id	54894
rvis_pct	86.32932295
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	colon_serrated_polyposis| sessile_serrated_polyposis_cancer_syndrome
gene_civic_url	.
description	.
---	---
RP11-763B22.9	
chrom	chr1
synonym	.
hgnc_id	.
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
RPL14	
chrom	chr3
synonym	L14,CTG-B33,hRL14,RL14
hgnc_id	10305
entrez_id	9045
rvis_pct	70.05779665
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
RPS6KB1	
chrom	chr17
synonym	p70(S6K)-alpha,PS6K,STK14A,S6K1
hgnc_id	10436
entrez_id	6198
rvis_pct	22.09247464

is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
RUNX2	
chrom	chr6
synonym	AML3,CBFA1,PEBP2A1,PEBP2aA1,CCD,CCD1
hgnc_id	10472
entrez_id	860
rvis_pct	24.33356924
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	cleidocranial_dysostosis| cleidocranial_dysplasia,forme_fruste,dental_anomalies_only| cleidocranial_dysplasia,forme_fruste,with_brachydactyly| cleidocranial_dysplasia,severe,with_osteoporosis_and_scoliosis
gene_civic_url	.
description	.
---	---
SDHA	
chrom	chr5
synonym	FP,SDH2,SDHF
hgnc_id	10680
entrez_id	6389
rvis_pct	10.95187544
is_OG	0
is_TS	1
in_cgi_biomarkers	.
clinvar_gene_phenotype	carney_triad| dilated_cardiomyopathy_1gg| hereditary_cancer-predisposing_syndrome| leigh_syndrome| mitochondrial_complex_ii_deficiency| paragangliomas_5| pheochromocytoma| skeletal_myopathy
gene_civic_url	.
description	.
---	---
SETD2	
chrom	chr3
synonym	KIAA1732,HIF-1,HYPB,FLJ23184,KMT3A
hgnc_id	18420
entrez_id	29072
rvis_pct	9.424392545
is_OG	0

is_TS	1
in_cgi_biomarkers	1
clinvar_gene_phenotype	luscan-lumish_syndrome
gene_civic_url	.
description	.
---	---
SMAD3	
chrom	chr15
synonym	MADH3,JV15-2,HsT17436
hgnc_id	6769
entrez_id	4088
rvis_pct	30.37272942
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	aneurysm| arterial_dissection| cutaneous_polyarteritis_nodosa| loeys-dietz_syndrome| loeys-dietz_syndrome_3| thoracic_aortic_aneurysm_and_aortic_dissection
gene_civic_url	.
description	.
---	---
SMARCA2	
chrom	chr9
synonym	Sth1p,SNF2L2,SNF2LA,BRM,BAF190,SNF2,hBRM,hSNF2a,SWI2
hgnc_id	11098
entrez_id	6595
rvis_pct	1.816466148
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	coffin_siris/intellectual_disability| nicolaides-baraitser_syndrome| severe_intellectual_deficiency
gene_civic_url	.
description	.
---	---
SMARCA4	
chrom	chr19
synonym	BRG1,SNF2L4,SNF2-BETA,SNF2LB,BAF190,FLJ39786,SNF2,hSNF2b,SWI2
hgnc_id	11100
entrez_id	6597
rvis_pct	0.601556971
is_OG	0
is_TS	0
in_cgi_biomarkers	1

clinvar_gene_phenotype	coffin-siris_syndrome| facial_asymmetry| global_developmental_delay| inborn_genetic_diseases| mental_retardation,autosomal_dominant_16| obesity| rhabdoid_tumor_predisposition_syndrome_2| single_transverse_palmar_crease| strabismus| ventricular_septal_defect| intellectual_deficiency
gene_civic_url	https://civic.genome.wustl.edu/links/genes/78
description	.
---	---
SNCAIP	
chrom	chr5
synonym	SYPH1
hgnc_id	11139
entrez_id	9627
rvis_pct	68.53621137
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	parkinson_disease,dominant/recessive| parkinson_disease,late-onset
gene_civic_url	.
description	.
---	---
SPEN	
chrom	chr1
synonym	KIAA0929,RBM15C,MINT,SHARP
hgnc_id	17575
entrez_id	23013
rvis_pct	1.209011559
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	ductal_breast_carcinoma
gene_civic_url	.
description	.
---	---
SPTA1	
chrom	chr1
synonym	EL2
hgnc_id	11272
entrez_id	6708
rvis_pct	99.79358339
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	elliptocytosis| elliptocytosis_2| hereditary_pyropoikilocytosis| spherocytosis,recessive| spherocytosis,type_3,autosomal_recessive| spherocytosis_type_3

gene_civic_url	.
description	.
---	---
SRC	
chrom	chr20
synonym	SRC1,c-src,ASV
hgnc_id	11283
entrez_id	6714
rvis_pct	14.96815287
is_OG	1
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	colon_cancer,advanced| thrombocytopenia_6
gene_civic_url	.
description	.
---	---
STAG2	
chrom	chrX
synonym	SA-2,SCC3B
hgnc_id	11355
entrez_id	10735
rvis_pct	17.03231894
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	None
gene_civic_url	https://civic.genome.wustl.edu/links/genes/8553
description	.
---	---
STAT4	
chrom	chr2
synonym	.
hgnc_id	11365
entrez_id	6775
rvis_pct	43.77211607
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	systemic_lupus_erythematosus_11
gene_civic_url	.
description	.
---	---

STAT5B

chrom	chr17
synonym	.
hgnc_id	11367
entrez_id	6777
rvis_pct	10.30313753
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	growth_hormone_insensitivity_with_immunodeficiency
gene_civic_url	.
description	.
---	---

SUZ12P

chrom	chr17
synonym	.
hgnc_id	.
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---

TAF1

chrom	chrX
synonym	BA2R,TAF2A,NSCL2,KAT4,DYT3/TAF1,DYT3,TAFII250,CCGS,CCG1
hgnc_id	11535
entrez_id	6872
rvis_pct	25.72540694
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	dystonia_3,torsion,x-linked| mental_retardation,x-linked,syndromic_33
gene_civic_url	.
description	.
---	---

TAF1L

chrom	chr9
synonym	.

hgnc_id	18056
entrez_id	138474
rvis_pct	17.75182826
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
TBP	
chrom	chr6
synonym	GTF2D1,SCA17,TFIID
hgnc_id	11588
entrez_id	6908
rvis_pct	36.86010852
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
TET1	
chrom	chr10
synonym	KIAA1676,CXXC6,LCX,bA119F7.1
hgnc_id	29484
entrez_id	80312
rvis_pct	76.31516867
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
TFE3	
chrom	chrX
synonym	bHLHe33,TFEA
hgnc_id	11752
entrez_id	7030
rvis_pct	80.00707714

is_OG	1
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
TGFBR2	
chrom	chr3
synonym	MFS2
hgnc_id	11773
entrez_id	7048
rvis_pct	50.34206181
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	aneurysm| ascending_aortic_aneurysm| congenital_aneurysm_of_ascending_aorta| hereditary_nonpolyposis_colorectal_cancer_type_6| loeys-dietz_syndrome| loeys-dietz_syndrome_1| loeys-dietz_syndrome_2| lynch_syndrome| malignant_tumor_of_esophagus| marfan_syndrome| thoracic_aortic_aneurysm_and_aortic_dissection
gene_civic_url	.
description	.
---	---
TMPRSS2	
chrom	chr21
synonym	PRSS10
hgnc_id	11876
entrez_id	7113
rvis_pct	72.75300778
is_OG	0
is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	malignant_tumor_of_prostate
gene_civic_url	https://civic.genome.wustl.edu/links/genes/5813
description	.
---	---
TOP2A	
chrom	chr17
synonym	TOP2
hgnc_id	11989
entrez_id	7153
rvis_pct	79.60603916
is_OG	0

is_TS	0
in_cgi_biomarkers	1
clinvar_gene_phenotype	dna_topoisomerase_ii,resistance_to_inhibition_of,by_amsacrine| long_qt_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/5848
description	.
---	---
TPRXL	
chrom	chr3
synonym	FLJ35107
hgnc_id	32178
entrez_id	.
rvis_pct	.
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
TSC1	
chrom	chr9
synonym	LAM,hamartin,TSC,KIAA0243
hgnc_id	12362
entrez_id	7248
rvis_pct	7.832035858
is_OG	0
is_TS	1
in_cgi_biomarkers	1
clinvar_gene_phenotype	adenoma_sebaceum| autism_spectrum_disorders| cardiac_rhabdomyoma| cortical_dysplasia| cortical_tubers| focal_cortical_dysplasia_of_taylor| focal_cortical_dysplasia_of_taylor_type_2b| hamartoma| hereditary_cancer-predisposing_syndrome| lymphangiomyomatosis| malignant_tumor_of_urinary_bladder| multiple_renal_cysts| renal_cortical_cysts| renal_insufficiency| seizures| tuberous_sclerosis_1| tuberous_sclerosis_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/46
description	.
---	---
TSC2	
chrom	chr16
synonym	LAM,TSC4,tuberin
hgnc_id	12363
entrez_id	7249
rvis_pct	0.87284737
is_OG	0
is_TS	1

in_cgi_biomarkers	1
clinvar_gene_phenotype	autism_spectrum_disorders| cortical_tubers| familial_adenomatous_polyposis_3| hereditary_cancer-predisposing_syndrome| hirschsprung_disease_1| infantile_spasms| intellectual_disability,severe| lymphangiomyomatosis| neoplasm_of_brain| tuberous_sclerosis_2| tuberous_sclerosis_and_lymphangiomyomatosis| tuberous_sclerosis_syndrome
gene_civic_url	https://civic.genome.wustl.edu/links/genes/47
description	.
---	---
TSPAN31	
chrom	chr12
synonym	SAS
hgnc_id	10539
entrez_id	6302
rvis_pct	65.33380514
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	cutaneous_malignant_melanoma,dominant| cutaneous_malignant_melanoma_3| hereditary_cancer-predisposing_syndrome| hereditary_cutaneous_melanoma
gene_civic_url	.
description	.
---	---
VEZF1	
chrom	chr17
synonym	DB1,ZNF161
hgnc_id	12949
entrez_id	7716
rvis_pct	16.35999056
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
XPO1	
chrom	chr2
synonym	CRM1,emb
hgnc_id	12825
entrez_id	7514
rvis_pct	11.05803255
is_OG	0
is_TS	0
in_cgi_biomarkers	.

clinvar_gene_phenotype	None
gene_civic_url	.
description	.
---	---
ZFHX3	
chrom	chr16
synonym	ZNF927,ATBF1
hgnc_id	777
entrez_id	463
rvis_pct	0.117952347
is_OG	0
is_TS	0
in_cgi_biomarkers	.
clinvar_gene_phenotype	inborn_genetic_diseases| prostate_cancer,somatic
gene_civic_url	.
description	.

Anhang B

Annotationsquellen

Dataset: Annotation Metadata

Contents:

Annotation	Version
dbSNP	b147.20160601
CancerHotspots	v2
CIViC variants	01-Apr-2021
CGI variants	13-Feb-2019
Uniprot cancer genes	13-Feb-2019
CIViC genes	01-Feb-2019
CGI genes	13-Feb-2019

Alle weiteren Annotationen basieren auf `SnpEff v4.3` und `Gemini v0.20.1`.

Protokollierung des verwendeten Workflows

Workflow: Panel_workflow_GEMINI_hg19_save

Steps:

Step	Annotation
Sample identifier	
reverse reads	
forward reads	
Annotations data	
Step 5	
Extract metadata	
Step 7	
Step 8	
Step 9	
Step 10	
Step 11	
Step 12	

Step	Annotation
Step 13	
Step 14	
Annotation metadata	
Step 16	
Step 17	
Step 18	
dbSNP annotations	
cancerhotspots data	
CIViC variant data	
CGI biomarkers	
UniProt-annotated cancer genes	
CIViC gene data	
CGI-listed genes	
Step 26	
Step 27	
Step 28	
canonical chromosomes	
Step 30	
Step 31	
Step 32	
Step 33	
Step 34	
Step 35	
Step 36	
Step 37	
Step 38	
Step 39	Add effects on genes, transcripts and proteins to variant INFO field
Step 40	
Step 41	Build GEMINI database of all variants
Step 42	
Step 43	Add somatic call stats
Step 44	Add selected dbsnp info

Step	Annotation
Step 45	Add cancerhotspots info
Step 46	Add CIViC link
Step 47	Add CGI Biomarkers info
GEMINI gene-centric query panel variants	
Generate helper report	
Generate basic MAF of panel variants	
GEMINI query panel variants	
Step 52	restore mal-formatted commas
Step 53	
Step 54	
Step 55	Add whitespace to comma- or pipe-separated multi-value fields (to allow for wrapping in final report)
Step 56	Add whitespace to pipe-separated multi-value fields (to allow for wrapping in final report)
Step 57	
Step 58	
Step 59	
Step 60	
Step 61	
Step 62	
Step 63	
Step 64	
Step 65	
Step 66	
Step 67	
Generate full gene report	
Get final MAF	
Step 70	

Step	Annotation
Step 71	The following "Split file" step doesn't emit the header line to the dataset created from the last line of input. We add an extra line to avoid malformatting and an empty collection when the tabular report consists of a header.
Step 72	
Step 73	
Step 74	
Step 75	
Step 76	
Step 77	
Generate full variants report	
Step 79	
Step 80	
Step 81	
Mutation Summary	
Filter for known cancer genes	
Step 84	
Step 85	
Step 86	
All variants report	
Variants report cancer genes	