

Patient Data	
Patient	DEF ABC
Birthdate	12.12.1212
Diagnosis	Melanoma

Mutation load	Medium	Number of non-synonymous SNVs	131
Number of oncogenes	3		
Number of tumor suppressor genes	6		
Additional information			

Somatic Mutations in Known Driver Genes

List of cancer driver genes along with the mutations observed in the patient. Confidence column shows the number of the driver gene sources that cataloged the corresponding gene as driver and Reference column gives the list of those sources.

Gene	Mutation	Driver Type	Confidence ¹	Reference
BRAF	p.Val600Glu	Oncogene	4	1,2,3,4
SF3B1	p.Pro718Leu	Oncogene	3	1,2,4
DLEC1	p.Asp215Asn	TSG	2	3,5
FAM46C	p.Thr209Asn	unknown	2	1,4
GLI1	p.Ser1094Phe	TSG/Oncogene	2	3,5
RPS6KA2	p.Glu319Lys	TSG	2	3,5
ACHE	p.Thr95Ile	TSG	1	5
EPHB4	p.Pro346Leu	TSG	1	5
ETV5	p.Tyr445Cys	unknown	1	1
LPP	p.Ala119Gly	unknown	1	1
MADD	p.Ser1620Phe	TSG	1	5
PABPC3	p.Gly234Arg	unknown	1	4
PCSK5	p.Cys747Tyr	unknown	1	4
TNPO1	p.Gln38His	unknown	1	4

¹ Confidence shows the number of driver gene sources that includes the gene. The sources are Vogelstein et al., Rubio-Perez et al., TSGene DB, COSMIC DB, UniProt.

Somatic Mutations with Known Pharmacogenetic Effect

List of drugs with the evidence of targeting the observed variant of the mutated gene regardless of the cancer type. The information is obtained from CIViC database. CIViC evidence levels are given in the Evidence column.

Gene	Mutation	Therapy	Effect	Disease	Evidence ²	References
BRAF	V600E	Bevacizumab	Resistance	Colorectal Cancer	B	31
BRAF	V600E	Cetuximab	Resistance	Colorectal Cancer	B	23
BRAF	V600E	Cetuximab, Vemurafenib, Irinotecan	Sensitivity/Response	Colorectal Cancer	B	30
BRAF	V600E	Dabrafenib, Trametinib	Sensitivity/Response	Melanoma	B	22
BRAF	V600E	Vemurafenib	Sensitivity/Response	Melanoma	B	25,29
BRAF	V600E	Vemurafenib	Sensitivity/Response	Ovarian Cancer	B	33
BRAF	V600E	Vemurafenib, Cobimetinib	Sensitivity/Response	Melanoma	B	27
BRAF	V600E	Dabrafenib	Resistance	Non-small Cell Lung Carcinoma	C	36
BRAF	V600E	Dabrafenib, Trametinib DMSO	Sensitivity/Response	Cholangiocarcinoma	C	52,53
BRAF	V600E	Pertuzumab, Vemurafenib	Sensitivity/Response	Anaplastic Thyroid Carcinoma	C	33
BRAF	V600E	Pictilisib	Sensitivity/Response	Melanoma	C	39
BRAF	V600E	Trametinib DMSO, Dabrafenib	Sensitivity/Response	Cholangiocarcinoma	C	54
BRAF	V600E	Vemurafenib	Sensitivity/Response	Colorectal Cancer	C	33
BRAF	V600E	Vemurafenib	Sensitivity/Response	Ganglioglioma	C	49
BRAF	V600E	Vemurafenib	Sensitivity/Response	Laryngeal Squamous Cell Carcinoma	C	33
BRAF	V600E	Vemurafenib	Sensitivity/Response	Multiple Myeloma	C	47,47
BRAF	V600E	Vemurafenib	Sensitivity/Response	Ovarian Cystadenocarcinoma	C	50
BRAF	V600E	Vemurafenib	Sensitivity/Response	Papillary Thyroid Carcinoma	C	42

² [CIViC evidence levels are used](#). A = Validated association, B = Clinical evidence, C = Case study, D = Preclinical evidence, E = Inferential association

Somatic Mutations with Known Pharmacogenetic Effect

List of drugs with the evidence of targeting the observed variant of the mutated gene regardless of the cancer type. The information is obtained from CIViC database. CIViC evidence levels are given in the Evidence column.

Gene	Mutation	Therapy	Effect	Disease	Evidence	References
BRAF	V600E	Vemurafenib, Panitumumab	Sensitivity/Response	Colorectal Cancer	C	44
BRAF	V600E	Vemurafenib, Panitumumab, Irinotecan	Sensitivity/Response	Cholangiocarcinoma	C	55
BRAF	V600E	BEZ235 (NVP-BEZ235, Dactolisib), GDC-0879	Sensitivity/Response	Colorectal Cancer	D	67
BRAF	V600E	Capecitabine, Vemurafenib, Bevacizumab	Sensitivity/Response	Colorectal Cancer	D	60
BRAF	V600E	Cobimetinib	Sensitivity/Response	Cancer	D	64
BRAF	V600E	PLX4720, GDC0941	Sensitivity/Response	Colorectal Cancer	D	58
BRAF	V600E	PLX4720, Nutlin-3	Sensitivity/Response	Colorectal Cancer	D	59
BRAF	V600E	Selumetinib (AZD6244), BEZ235 (NVP-BEZ235, Dactolisib)	Sensitivity/Response	Melanoma	D	63
BRAF	V600E	Sorafenib, Panitumumab	Sensitivity/Response	Colorectal Cancer	D	56
BRAF	V600E	Vemurafenib	Resistance	Melanoma	D	57
BRAF	V600E	Vemurafenib	Sensitivity/Response	Colorectal Cancer	D	60
BRAF	V600E	Vemurafenib, Gefitinib, Cetuximab	Sensitivity/Response	Colorectal Cancer	D	66

Somatic Mutations in Pharmaceutical Target proteins

CIViC Summary of Drugs Targeting Affected Genes

Therapies that have evidence of targeting the affected gene. The information is obtained from CIViC database. CIViC evidence levels are given in Evidence column. Results are filtered according to cancer type, if it is provided in metadata.

Gene	Mutation	Therapy	Effect	Disease	Evidence ³	References
BRAF	L505H	Vemurafenib	Resistance	Melanoma	B	28
BRAF	V600	Dabrafenib	Sensitivity/Response	Melanoma	B	26
BRAF	V600	RO4987655	Sensitivity/Response	Melanoma	B	24
BRAF	V600D	Dabrafenib	Sensitivity/Response	Melanoma	B	21
BRAF	V600E	Dabrafenib, Trametinib	Sensitivity/Response	Melanoma	B	22
BRAF	V600E	Vemurafenib	Sensitivity/Response	Melanoma	B	25,29
BRAF	V600E	Vemurafenib, Cobimetinib	Sensitivity/Response	Melanoma	B	27
BRAF	V600K	Dabrafenib, Trametinib	Sensitivity/Response	Melanoma	B	27
BRAF	V600K	Vemurafenib	Sensitivity/Response	Melanoma	B	25
BRAF	AGK-BRAF	Sorafenib	Sensitivity/Response	Melanoma	C	37
BRAF	L597R	Vemurafenib	Sensitivity/Response	Melanoma	C	38
BRAF	V600	BAY 86-9766	Resistance	Melanoma	C	40
BRAF	V600E	Pictilisib	Sensitivity/Response	Melanoma	C	39
BRAF	V600E+V600M	Dabrafenib	Sensitivity/Response	Melanoma	C	35
BRAF	V600K	Vemurafenib	Sensitivity/Response	Melanoma	C	41
BRAF	AGK-BRAF	Vemurafenib	Resistance	Melanoma	D	37
BRAF	DEL 485-490	LY3009120	Sensitivity/Response	Cancer	D	65
BRAF	L505H	Vemurafenib	Resistance	Melanoma	D	69
BRAF	MUTATION	Trametinib	Sensitivity/Response	Cancer	D	62
BRAF	PAPSS1-BRAF	Trametinib	Sensitivity/Response	Melanoma	D	61
BRAF	PAPSS1-BRAF	Vemurafenib	Resistance	Melanoma	D	61
BRAF	TRIM24-BRAF	Trametinib	Sensitivity/Response	Melanoma	D	61
BRAF	V600D	Vemurafenib	Sensitivity/Response	Melanoma	D	72,73
BRAF	V600E	Cobimetinib	Sensitivity/Response	Cancer	D	64
BRAF	V600E	Selumetinib	Sensitivity/Response	Melanoma	D	63

³ [CIViC evidence levels are used](#). A = Validated association, B = Clinical evidence, C = Case study, D = Preclinical evidence, E = Inferential association

Somatic Mutations in Pharmaceutical Target proteins

CIViC Summary of Drugs Targeting Affected Genes

Therapies that have evidence of targeting the affected gene. The information is obtained from CIViC database. CIViC evidence levels are given in Evidence column. Results are filtered according to cancer type, if it is provided in metadata.

Gene	Mutation	Therapy	Effect	Disease	Evidence	References
BRAF	V600E	(AZD6244),BEZ235 (NVP-BEZ235, Dactolisib)	Resistance	Melanoma	D	57

Summary of Cancer Drugs Targeting Affected Genes

List of cancer drugs targeting the mutated gene. Information is obtained from DrugBank, Therapeutic Target Database, IUPHAR, and Santos et al.

Gene	Status	Therapy	Confidence ⁴	References
BRAF	approved investigational	sorafenib	9	6,7,8,9,10,11,12,13,14
BRAF	approved	dabrafenib	4	12,14,15
BRAF	approved	vemurafenib	4	12,14,16,17
BRAF	approved	regorafenib	3	12,18
TNFRSF8	approved	brentuximab vedotin	2	12,19
BRAF	approved	gsk2118436	1	14
BRAF	approved	r7204	1	14
EPHB4	approved	vandetanib	1	12

References

The publications of the reference IDs given in the tables above.

1	Futreal et al., A census of human cancer genes., Nature reviews. Cancer, 4, 3, 2004
2	Vogelstein et al., Cancer genome landscapes., Science (New York, N.Y.), 339, 6127, 2013
3	Apweiler et al., UniProt: the Universal Protein knowledgebase., Nucleic acids research, 32, Database issue, 2004
4	Rubio et al., In silico prescription of anticancer drugs to cohorts of 28 tumor types reveals targeting opportunities., Cancer cell, 27, 3, 2015
5	Zhao et al., TSGene: a web resource for tumor suppressor genes., Nucleic acids research, 41, Database issue, 2013
6	Flaherty et al., Chemotherapy and targeted therapy combinations in advanced melanoma., Clinical cancer research : an official journal of the American Association for Cancer Research, 12, 7 Pt 2, 2006
7	Haluska et al., Therapeutic targets in melanoma: map kinase pathway., Current oncology reports, 8, 5, 2006
8	Kim et al., Sorafenib inhibits the angiogenesis and growth of orthotopic anaplastic thyroid

4 Confidence shows the total number of the publications supporting the association.

References

	carcinoma xenografts in nude mice., Molecular cancer therapeutics, 6, 6, 2007
9	Eisen et al., Sorafenib in advanced melanoma: a Phase II randomised discontinuation trial analysis., British journal of cancer, 95, 5, 2006
10	Lu et al., Sorafenib induces growth inhibition and apoptosis of human chondrosarcoma cells by blocking the RAF/ERK/MEK pathway., Journal of surgical oncology, 102, 7, 2010
11	Chen et al., TTD: Therapeutic Target Database., Nucleic acids research, 30, 1, 2002
12	Santos et al., A comprehensive map of molecular drug targets., Nature reviews. Drug discovery, 16, 1, 2017
13	Wilhelm et al., BAY 43-9006 exhibits broad spectrum oral antitumor activity and targets the RAF/MEK/ERK pathway and receptor tyrosine kinases involved in tumor progression and angiogenesis., Cancer research, 64, 19, 2004
15	Gibney et al., Clinical development of dabrafenib in BRAF mutant melanoma and other malignancies., Expert opinion on drug metabolism & toxicology, 9, 7, 2013
16	Jordan et al., Vemurafenib for the treatment of melanoma., Expert opinion on pharmacotherapy, 13, 17, 2012
17	Wang et al., Conformation-specific effects of Raf kinase inhibitors., Journal of medicinal chemistry, 55, 17, 2012
18	Zambon et al., Small molecule inhibitors of BRAF in clinical trials., Bioorganic & medicinal chemistry letters, 22, 2, 2012
19	Francisco et al., cAC10-vcMMAE, an anti-CD30-monomethyl auristatin E conjugate with potent and selective antitumor activity., Blood, 102, 4, 2003
20	Peeters et al., Massively parallel tumor multigene sequencing to evaluate response to panitumumab in a randomized phase III study of metastatic colorectal cancer., Clinical cancer research : an official journal of the American Association for Cancer Research, 19, 7, 2013
21	Ponti et al., The somatic affairs of BRAF: tailored therapies for advanced malignant melanoma and orphan non-V600E (V600R-M) mutations., Journal of clinical pathology, 66, 5, 2013
22	Menzies et al., Dabrafenib and trametinib, alone and in combination for BRAF-mutant metastatic melanoma., Clinical cancer research : an official journal of the American Association for Cancer Research, 20, 8, 2014
23	De et al., Effects of KRAS, BRAF, NRAS, and PIK3CA mutations on the efficacy of cetuximab plus chemotherapy in chemotherapy-refractory metastatic colorectal cancer: a retrospective consortium analysis., The Lancet. Oncology, 11, 8, 2010
24	Zimmer et al., Phase I expansion and pharmacodynamic study of the oral MEK inhibitor RO4987655 (CH4987655) in selected patients with advanced cancer with RAS-RAF mutations., Clinical cancer research : an official journal of the American Association for Cancer Research, 20, 16, 2014
25	McArthur et al., Safety and efficacy of vemurafenib in BRAF(V600E) and BRAF(V600K) mutation-positive melanoma (BRIM-3): extended follow-up of a phase 3, randomised, open-label study., The Lancet. Oncology, 15, 3, 2014
26	Falchook et al., Dabrafenib in patients with melanoma, untreated brain metastases, and other solid tumours: a phase 1 dose-escalation trial., Lancet (London, England), 379, 9829, 2012
27	Larkin et al., Combined vemurafenib and cobimetinib in BRAF-mutated melanoma., The New England journal of medicine, 371, 20, 2014
28	Hoogstraat et al., Detailed imaging and genetic analysis reveal a secondary BRAF(L505H) resistance mutation and extensive inpatient heterogeneity in metastatic BRAF mutant melanoma patients treated with vemurafenib., Pigment cell & melanoma research, 28, 3, 2015
29	Flaherty et al., Inhibition of mutated, activated BRAF in metastatic melanoma., The New England

References

	journal of medicine, 363, 9, 2010
30	Hong et al., Phase IB Study of Vemurafenib in Combination with Irinotecan and Cetuximab in Patients with Metastatic Colorectal Cancer with BRAFV600E Mutation., Cancer discovery, 6, 12, 2016
31	Tol et al., BRAF mutation in metastatic colorectal cancer., The New England journal of medicine, 361, 1, 2009
32	Hyman et al., Vemurafenib in Multiple Nonmelanoma Cancers with BRAF V600 Mutations., The New England journal of medicine, 373, 8, 2015
33	Hainsworth et al., Targeted Therapy for Advanced Solid Tumors on the Basis of Molecular Profiles: Results From MyPathway, an Open-Label, Phase IIa Multiple Basket Study., Journal of clinical oncology : official journal of the American Society of Clinical Oncology, 36, 6, 2018
34	Souglakos et al., Prognostic and predictive value of common mutations for treatment response and survival in patients with metastatic colorectal cancer., British journal of cancer, 101, 3, 2009
35	Ponti et al., Overwhelming response to Dabrafenib in a patient with double BRAF mutation (V600E; V600M) metastatic malignant melanoma., Journal of hematology & oncology, 5, , 2012
36	Rudin et al., Molecular characterization of acquired resistance to the BRAF inhibitor dabrafenib in a patient with BRAF-mutant non-small-cell lung cancer., Journal of thoracic oncology : official publication of the International Association for the Study of Lung Cancer, 8, 5, 2013
37	Botton et al., Recurrent BRAF kinase fusions in melanocytic tumors offer an opportunity for targeted therapy., Pigment cell & melanoma research, 26, 6, 2013
38	Bahadoran et al., Major clinical response to a BRAF inhibitor in a patient with a BRAF L597R-mutated melanoma., Journal of clinical oncology : official journal of the American Society of Clinical Oncology, 31, 19, 2013
39	Sarker et al., First-in-human phase I study of pictilisib (GDC-0941), a potent pan-class I phosphatidylinositol-3-kinase (PI3K) inhibitor, in patients with advanced solid tumors., Clinical cancer research : an official journal of the American Association for Cancer Research, 21, 1, 2015
40	Weekes et al., Multicenter phase I trial of the mitogen-activated protein kinase 1/2 inhibitor BAY 86-9766 in patients with advanced cancer., Clinical cancer research : an official journal of the American Association for Cancer Research, 19, 5, 2013
41	Sahadudheen et al., Long Term Survival and Continued Complete Response of Vemurafenib in a Metastatic Melanoma Patient with BRAF V600K Mutation., Case reports in oncological medicine, 2016, , 2016
42	Ali et al., Extended Antitumor Response of a BRAF V600E Papillary Thyroid Carcinoma to Vemurafenib., Case reports in oncology, 7, 2, 2014
43	Menzies et al., Clinical activity of the MEK inhibitor trametinib in metastatic melanoma containing BRAF kinase fusion., Pigment cell & melanoma research, 28, 5, 2015
44	Pietrantonio et al., MET-Driven Resistance to Dual EGFR and BRAF Blockade May Be Overcome by Switching from EGFR to MET Inhibition in BRAF-Mutated Colorectal Cancer., Cancer discovery, 6, 9, 2016
45	Grisham et al., Extreme Outlier Analysis Identifies Occult Mitogen-Activated Protein Kinase Pathway Mutations in Patients With Low-Grade Serous Ovarian Cancer., Journal of clinical oncology : official journal of the American Society of Clinical Oncology, 33, 34, 2015
46	Subbiah et al., Targeted therapy by combined inhibition of the RAF and mTOR kinases in malignant spindle cell neoplasm harboring the KIAA1549-BRAF fusion protein., Journal of hematology & oncology, 7, , 2014
47	Sharman et al., Vemurafenib response in 2 patients with posttransplant refractory BRAF V600E-mutated multiple myeloma., Clinical lymphoma, myeloma & leukemia, 14, 5, 2014

References

48	Ahronian et al., Clinical Acquired Resistance to RAF Inhibitor Combinations in BRAF-Mutant Colorectal Cancer through MAPK Pathway Alterations., <i>Cancer discovery</i> , 5, 4, 2015
49	del et al., Response of recurrent BRAFV600E mutated ganglioglioma to Vemurafenib as single agent., <i>Journal of translational medicine</i> , 12, , 2014
50	Combe et al., Sustained response to vemurafenib in a low grade serous ovarian cancer with a BRAF V600E mutation., <i>Investigational new drugs</i> , 33, 6, 2015
51	Hsu et al., Mutations of KRAS/NRAS/BRAF predict cetuximab resistance in metastatic colorectal cancer patients., <i>Oncotarget</i> , 7, 16, 2016
52	Kocsis et al., Combined dabrafenib and trametinib treatment in a case of chemotherapy-refractory extrahepatic BRAF V600E mutant cholangiocarcinoma: dramatic clinical and radiological response with a confusing synchronic new liver lesion., <i>Journal of gastrointestinal oncology</i> , 8, 2, 2017
53	Lavingia et al., Impressive response to dual &BRAF& and MEK inhibition in patients with BRAF mutant intrahepatic cholangiocarcinoma-2 case reports and a brief review., <i>Journal of gastrointestinal oncology</i> , 7, 6, 2016
54	Loaiza et al., Dramatic response to dabrafenib and trametinib combination in a BRAF V600E-mutated cholangiocarcinoma: implementation of a molecular tumour board and next-generation sequencing for personalized medicine., <i>Ecancermedicalscience</i> , 8, , 2014
55	Silkin et al., Complete Clinical Response of BRAF-Mutated Cholangiocarcinoma to Vemurafenib, Panitumumab, and Irinotecan., <i>Journal of gastrointestinal cancer</i> , 47, 4, 2016
56	Di et al., Wild-type BRAF is required for response to panitumumab or cetuximab in metastatic colorectal cancer., <i>Journal of clinical oncology : official journal of the American Society of Clinical Oncology</i> , 26, 35, 2008
57	Nissan et al., Loss of NF1 in cutaneous melanoma is associated with RAS activation and MEK dependence., <i>Cancer research</i> , 74, 8, 2014
58	Rad et al., A genetic progression model of Braf(V600E)-induced intestinal tumorigenesis reveals targets for therapeutic intervention., <i>Cancer cell</i> , 24, 1, 2013
59	Ji et al., Vemurafenib synergizes with nutlin-3 to deplete survivin and suppresses melanoma viability and tumor growth., <i>Clinical cancer research : an official journal of the American Association for Cancer Research</i> , 19, 16, 2013
60	Yang et al., Antitumor activity of BRAF inhibitor vemurafenib in preclinical models of BRAF-mutant colorectal cancer., <i>Cancer research</i> , 72, 3, 2012
61	Hutchinson et al., BRAF fusions define a distinct molecular subset of melanomas with potential sensitivity to MEK inhibition., <i>Clinical cancer research : an official journal of the American Association for Cancer Research</i> , 19, 24, 2013
62	Jing et al., Comprehensive predictive biomarker analysis for MEK inhibitor GSK1120212., <i>Molecular cancer therapeutics</i> , 11, 3, 2012
63	Penna et al., Primary cross-resistance to BRAFV600E-, MEK1/2- and PI3K/mTOR-specific inhibitors in BRAF-mutant melanoma cells counteracted by dual pathway blockade., <i>Oncotarget</i> , 7, 4, 2016
64	Hatzivassiliou et al., Mechanism of MEK inhibition determines efficacy in mutant KRAS- versus BRAF-driven cancers., <i>Nature</i> , 501, 7466, 2013
65	Chen et al., Oncogenic BRAF Deletions That Function as Homodimers and Are Sensitive to Inhibition by RAF Dimer Inhibitor LY3009120., <i>Cancer discovery</i> , 6, 3, 2016
66	Prahallad et al., Unresponsiveness of colon cancer to BRAF(V600E) inhibition through feedback activation of EGFR., <i>Nature</i> , 483, 7387, 2012
67	Coffee et al., Concomitant BRAF and PI3K/mTOR blockade is required for effective treatment of BRAF(V600E) colorectal cancer., <i>Clinical cancer research : an official journal of the American Association for Cancer Research</i> , 19, 10, 2013

References

68	Dahlman et al., BRAF(L597) mutations in melanoma are associated with sensitivity to MEK inhibitors., Cancer discovery, 2, 9, 2012
69	Wagenaar et al., Resistance to vemurafenib resulting from a novel mutation in the BRAFV600E kinase domain., Pigment cell & melanoma research, 27, 1, 2014
70	Bertotti et al., A molecularly annotated platform of patient-derived xenografts ("xenopatients") identifies HER2 as an effective therapeutic target in cetuximab-resistant colorectal cancer., Cancer discovery, 1, 6, 2011
71	NA et al., NA, NA, NA, NA, NA, NA
72	Tsai et al., Discovery of a selective inhibitor of oncogenic B-Raf kinase with potent antimelanoma activity., Proceedings of the National Academy of Sciences of the United States of America, 105, 8, 2008
73	Yang et al., RG7204 (PLX4032), a selective BRAFV600E inhibitor, displays potent antitumor activity in preclinical melanoma models., Cancer research, 70, 13, 2010
74	Smalley et al., CRAF inhibition induces apoptosis in melanoma cells with non-V600E BRAF mutations., Oncogene, 28, 1, 2009
75	Corcoran et al., BRAF gene amplification can promote acquired resistance to MEK inhibitors in cancer cells harboring the BRAF V600E mutation., Science signaling, 3, 149, 2010
76	Heidorn et al., Kinase-dead BRAF and oncogenic RAS cooperate to drive tumor progression through CRAF., Cell, 140, 2, 2010

Appendix

All the somatic variants of the patient with their dbSNP and COSMIC IDs.

Gene	Mutation	dbSNP	COSMIC
TNFRSF8	p.Pro215Ser	rs267597959	COSM14024
FAM46C	p.Thr209Asn		
S100A7A	p.Gly98Trp	rs267598049	COSM36721
PKLR	p.Gly251Ser	rs267598065	COSM36782
MAEL	p.Ser431Cys	rs267598149	COSM36684
ZBTB41	p.Phe164Val	rs267598277	COSM36692
SYT14	p.Ser437Phe	rs267598356	COSM36786
OR2T8	p.Met197Arg	rs4474294	
OR2T3	p.Ala214Thr	rs1770109	
SLC4A5	p.Ser428Phe	rs111392973	COSM2999241,COSM2999242
SLC4A5	p.Ser428Thr	rs267599454	
SNRNP200	p.Arg1538Cys	rs267599495	COSM36589
SEMA4C	p.Arg407Trp	rs267599501	COSM36666
ANKRD36	p.Ser1120Cys	rs768768868	
KIAA1211L	p.Gly746Glu	rs866719486	
DPP10	p.Ile93Asn		
XIRP2	p.Gly127Arg	rs267598980	COSM36673

Appendix			
TTN	p.Pro10904Ser	rs267599054	
TTN	p.Ala1347Thr	rs267599092	COSM2708938,COSM2708939,COSM2708940,COSM2708941,COSM2708942
SF3B1	p.Pro718Leu	rs267599150	COSM36655
FZD7	p.Pro285Ser	rs267599158	COSM24315
ZDBF2	p.Gly575Arg		
AGFG1	p.Gly364Arg	rs267599235	COSM25632,COSM3364621,COSM3364622
ARL4C	p.Gly71Ser	rs61752230	COSM21657
KIF1A	p.Ser141Ala		
DLEC1	p.Asp215Asn	rs149190717	COSM1566798,COSM36702
VPRBP	p.Pro309Leu	rs267599884	
TLR9	p.Gly514Ser	rs267599888	COSM36649
PRR23C	p.Glu262Lys	rs759730911	COSM36858
CLSTN2	p.Gln262His	rs267599628	COSM36631
SAMD7	p.Arg67Trp	rs191885635	COSM36663
GNB4	p.Pro107Leu	rs267599699	COSM13667
ETV5	p.Tyr445Cys	rs267599722	COSM23333
LPP	p.Ala119Gly		
MUC4	p.Pro1056His	rs753583962	
EXOC1	p.Pro774Ser	rs267600192	COSM36662
REST	p.Pro752Thr	rs267600197	COSM24349
SMR3B	p.Arg58Lys	rs267600235	COSM1310225,COSM36745
ADAM29	p.Gly589Glu	rs267600094	COSM26290
CARD6	p.Leu638Phe	rs267600630	COSM14006
TNPO1	p.Gln38His	rs267600680	COSM36775,COSM5648790
F2RL2	p.Leu141Phe	rs267600693	COSM27249,COSM36780
VCAN	p.Asp203Asn	rs267600718	COSM36758
EDIL3	p.Gln187Lys	rs267600722	COSM26295
PCDHB7	p.Asp374His		
GRIA1	p.Gly828Glu	rs267600500	COSM36714,COSM4854071,COSM4854072
ADAM19	p.Pro900Leu	rs61757467	
MBOAT1	p.Lys293Asn		
SPDEF	p.Asp283His		

Appendix			
SPDEF	p.Ser229Leu	rs200344679	COSM36760
SCUBE3	p.Gly702Glu	rs267600995	COSM36691
BTBD9	p.Arg46Cys	rs267601008	COSM36793
GPR111	p.Ile290Leu	rs267601055	COSM36619
BAI3	p.Asp755Asn	rs267601102	COSM22119
KATNA1	p.Pro241Leu	rs267600852	
KATNA1	p.Pro241Ser	rs267600853	
RPS6KA2	p.Glu319Lys	rs267600891	COSM21036,COSM3024932,COSM3024933
ANLN	p.Gln649Arg	rs267601502	COSM36632
ABCA13	p.Gly4948Asp	rs267601533	COSM36817
EPHB4	p.Pro346Leu	rs267601191	COSM21032
ACHE	p.Thr95Ile	rs267601193	COSM36706
BRAF	p.Val600Glu	rs113488022	COSM18443,COSM476,COSM6137
RP11-1220K2.2	p.Asp1426Glu		
TRBV23-1	p.Pro27Leu		COSM36861
ZNF862	p.Gln583Lys	rs267601404	COSM36833
NAT2	p.Glu264Lys	rs267601842	COSM36677
SCARA5	p.Glu270Lys	rs267601883	COSM36713
GPR124	p.Glu863Lys	rs267601912	COSM36641
REXO1L1P	p.Ser639Phe		
CNBD1	p.Leu135Arg		
GRHL2	p.Ser356Phe	rs267601682	COSM36601
ZC3H3	p.Ser879Phe	rs267601811	COSM36642
ANKRD18A	p.Glu654Lys	rs267602244	COSM36859
PCSK5	p.Cys747Tyr	rs267602276	COSM36640
NUTM2G	p.Gly36Asp	rs267602327	COSM36612
OR1J1	p.Leu157Phe	rs267602118	COSM36710
GAPVD1	p.Leu35Phe	rs267602131	COSM36617
ADAMTS13	p.Arg398His	rs121908471	COSM36777
LHX3	p.Gly92Glu		COSM36599
MADD	p.Ser1620Phe	rs267602903	COSM26934
OR4S2	p.Arg120Cys	rs267602971	COSM36685
OR4D11	p.Pro58Ala	rs267603040	COSM36624
SPTBN2	p.Glu2047Lys	rs201985455	COSM36751,COSM419893
GRM5	p.Glu941Lys	rs267603229	

Appendix			
DCP1B	p.Pro98Ser	rs267603408	COSM36575
CD163	p.Pro310Leu	rs267603681	COSM36725
GLI1	p.Ser1094Phe	rs267603606	COSM24658
TBC1D30	p.Gly327Glu	rs267603627	COSM36841
KCNC2	p.Leu298Ser	rs267603669	COSM36754
PABPC3	p.Gly234Arg	rs267603790	COSM36646
CPB2	p.Phe409Ser	rs267603833	COSM36708
CARKD	p.Pro205Ser	rs267603758	COSM36577
NOVA1	p.Ala256Asp	rs267603974	COSM1369439,COSM25331
ARHGAP5	p.Thr437Ile	rs56259828	
SERPINA6	p.Arg282Leu	rs267604111	COSM1265285,COSM26307,COSM267404
NUDT14	p.Thr44Pro	rs267603899	COSM36696
IGHV1-18	p.Gln20Lys		
TRPM1	p.Glu1261Lys	rs267604151	COSM36625
PLIN1	p.Leu191Arg		COSM36595
PRC1	p.Gly507Glu	rs267604387	COSM36743
AMDHD2	p.His587Arg		
NLRC3	p.Gly454Arg	rs267604538	COSM36804
TTLL6	p.Arg280Lys	rs267604932	COSM36852
USH1G	p.Leu379Ser	rs267605044	COSM36661
PSMA8	p.Gly36Glu	rs267605136	COSM36586
MBD3	p.Asp283Asn	rs369581342	
GTF2F1	p.Gly411Arg		
MUC16	p.Met2821Ile	rs267605807	COSM2701120,COSM36853
MUC16	p.Leu2819Met		
MUC16	p.Leu1434Ile		
OLFM2	p.Arg58Gln	rs267605828	COSM36654
PKN1	p.Arg191Cys	rs267605306	COSM21035
CYP4F2	p.Arg149Gln	rs140630977	COSM1129961
ZNF208	p.His855Tyr	rs267605385	
ARHGAP33	p.Pro1068Leu		
NOSIP	p.Pro297Leu		
ZNF880	p.Pro169Gln	rs267605631	COSM1234752
ZSCAN5A	p.Ala179Thr		
FLRT3	p.Ile532Asn		

Appendix			
DLGAP4	p.Ala879Ser	rs267605913	COSM36648
SPO11	p.Gly88Ser	rs267606012	COSM36690
TMEM50B	p.Ser113Phe	rs267606110	COSM36665
CECR2	p.Gly474Arg	rs267606173	COSM36851
IGLV3-12	p.Ala89Thr	rs2073451	
IGLJ3	p.Pro10Ala	rs2009433	
MEI1	p.Gly507Glu	rs267606261	COSM36800
NHS	p.Arg373Gln	rs267606412	COSM1118631,COSM36761
FGD1	p.Arg636Trp		COSM21850
PJA1	p.His586Tyr	rs267606501	
OGT	p.Leu367Ser		
DCAF12L1	p.Ser281Phe	rs267606338	COSM36778

Disclaimer

This report is intended as a hypothesis generating framework and is thus intended for research use only and not for diagnostic or clinical purposes. Information provided in this report does not replace a physician's medical judgement and usage is entirely at your own risk. The providers of this resource shall in no event be liable for any direct, indirect, incidental, consequential, or exemplary damages.