MTB Report

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PATIENT INFORMATION

Patient ID		**Tissue Type**	
Gender		**Tumor Content (%)**	
Disease	unspecified	**Number SNVs**	5408
Stage			
Previous Therapies			

GENE-DRUG PREDICTIVE ASSOCIATIONS

Method: Somatic variants of the patient (mutations, amplifications, deletions, rearrangments) are searched in curated databases of predictive biomarkers (GKDB[1], CIViC[2]) and reported according to their clinical evidence (see Levels of Evidence). In the following two tables, basic information of the somatic variants with relevant clinical implications can be found:

Gene	Patient's Variant	Level of Evidence
ABCB1	S1022R, S958R	B2
BRCA2	P360S, P237S, S1676I, I2672T, *194Q, I161T, Q2894R, Q383R	B1 ,B2
CDKN2A	G63A	B2 ,B3
DDR1	I1284V	unknown
DNMT3A	E18D	B1 ,B2
ERBB2	N133D, K62R, A733T, A748T, A487T, A763T	B2 ,B3
ERBB3	P453A, P512A, P31A	B2 ,B3
FANCA	C188S	B2
FAT1	D3429N	B3
FCGR2A	I266N, I226N, I181N, I267N, I128N, L19M	B2
FGFR3	M727T, M725T, M721T, M613T	B1 ,B2 ,B3
GNA11	T257S	B2 ,B3
IL7R	E172K, C390G	B3
MTAP	C55*, C72*	B3
MTOR	L2P, W1719R, W1790R, G279R	B2 ,B3
NF1	G1498W, G1513W, G1519W, G1529W, G1532W, G1164W, G115W	B2 ,B3
NF2	Q115*, Q73*, Q86*	B2 ,B3
NOTCH3	T784S	unknown
PTPRD	T781A	B2 ,B3
RAF1	L551R, W209G, L584R, L604R, L509R, L580R, L547R, L525R	B3
RIT1	Q116H, Q117H, Q80H, Q133H	B3
TSC1	L912Q, L861Q, L477Q, L906Q, L911Q, L907Q, E887K, E836K, E452K, E881K, E886K, E882K, A734T, A683T, A313T, A728T, A733T, A729T, M88I	B2
TSC2	G10C, G21C, Q7H, R1680L, R1724L, R1634L, R1749L, R1705L, R1682L, R1681L, R1747L, R1646L, R1726L, R1711L, R1683L, R1706L, R1702L, R1693L, G1723C	B2 ,B3
VHL	C176F	B2

Levels of Evidence: Findings are classified into 6 levels of evidence combining the **axis A-B** and the **axis 1-2-3**. Level A means evidence in the same cancer type. Level B means evidence in any other cancer type. On the 1-2-3 axis, level 1 means evidence supported by drug approval organizations or clinical guidelines, level 2 contains clinical evidence (clinical trials, case reports) and level 3 consists of preclinical evidence

Table of Results: All the predictive associations are detailed in this table. The results are sorted by 1) drug frequency, 2) levels of evidence (A1-B1-A2-B2-A3-B3). To allow a quick interpretation, the type of association (response, resistance) is colored (green, red) and new variants are gray.

Patient				Gene-Drug Associations				
Gene	**Variant**	**Disease**	**Known Variant**	**Predicts**	**Drugs**	**Evidence**	**PMID**	**Level**

[1] Dienstmann et al., Cancer Discov (2015), gdkd
[2] Griffith et al., Nat Genet (2017), civic

Patient				Gene-Drug	Associations			
Gene	**Variant**	**Disease**	**Known Variant**	**Predicts**	**Drugs**	**Evidence**	**PMID**	**Level**
ERBB2	N133D, K62R, A733T, A748T, A487T, A763T	lung	A775-G776insYVMA, G776delinsVC, V659E, S310F (GoF)	response	Ado-trastuzumab Emtansine	early trials	ASCO 2017 (abstr 8510)	**B2**
NF2	Q115*, Q73*, Q86*	breast	any variant (LoF)	response	Tensirolimus Plus Chemotherapy	case report	25878190	**B2**
TSC1	L912Q, L861Q, L477Q, L906Q, L911Q, L907Q, E887K, E836K, E452K, E881K, E886K, E882K, A734T, A683T, A313T, A728T, A733T, A729T, M88I	endometrial	any variant (GoF)	response	Tensirolimus	case report	27016228	**B2**
TSC2	G10C, G21C, Q7H, R1680L, R1724L, R1634L, R1749L, R1705L, R1682L, R1681L, R1747L, R1646L, R1726L, R1711L, R1683L, R1706L, R1702L, R1693L, G1723C	endometrial	any variant (GoF)	response	Tensirolimus	case report	27016228	**B2**

Other genes: here you can find other genes that might be interesting to check (information from Target DB[3] and Meric-Bernstam list[4]). No level information is provided in this section.

Patient		Drug-Gene Interactions		
Gene	**Variant**	**Known Variant**	**Description**	**Drugs**
DDR1	I1284V	Mutation; Amplification		Treatment with DDR1 inhibitor
NOTCH3	T784S	Mutation; Amplification; Rearrangement		Treatment with GSIs

[3] Van Allen et al., Nature medicine 20.6 (2014): 682-688, v3
[4] Meric-Bernstam et al., J Natl Cancer Inst. 107(7) (2015)