参考信息基因列表共检测基因117个其中：靶向药物相关基因117个，化疗/内分泌药物相关基因117个，HRD-DDR相关基因1个，肿瘤遗传易感相关基因1个，免疫治疗相关基因1个，评估微卫星状态的串联重复序列240个，用于评价肿瘤突变负荷/肿瘤新抗原的基因572个。其中：对1个基因报告单碱基和小片段基因变异，对1个基因报告基因重排，对1个基因报告拷贝数变异，对33个基因报告基因多态性。用于评价肿瘤突变负荷/肿瘤新抗原的基因572个。具体列表如下：以检测目的分类靶向药物相关基因117个ABL1DNMT3AINSRPIK3CAAKT1EGFRITKPIK3CDALKEPHA2JAK1PIK3R1ARERBB2JAK2PLCG2ARAFERBB3JAK3PTCH1AXLERBB4KDRPTENBAP1ESR1KITPTK6BLKFGFR1KRASPTPN11BRAFFGFR2LCKRAF1BRCA1FGFR3LYNRETBRCA2FGFR4MAP2K1ROS1BTKFLT1MAP2K2SMAD4CCND1FLT3MAP3K8SMOCD274FLT4METSRCCDK4FRKMTORSTK11CDK6GNA11MYCNTEKCDKN2AGNAQNF1TNFSF11CRLF2HCKNRASTNK2CSF1RHRASNTRK1TP53CTLA4IDH1PDGFRATSC1CTNNB1IDH2PDGFRBTSC2DDR1IGF1RPGRVEGFADDR2ABCB1ATICCBR3CDACYP3A4CYP2C8CYP2C19CYP2D6CYP19A1C8orf34DPYDERCC1ERCC2GSTP1MTHFRMTRRNQO1RRM1SLIT1SOD2TP53TPMTTYMSUGT1A1UGT1A9UMPSXRCC1XPC免疫治疗相关基因1个HR-DDR相关基因1个肿瘤遗传易感相关基因1个化疗/内分泌药物相关基因1个以检测范围分类报告重排（融合）的基因1个报告拷贝数变异的基因1个报告基因多态性的基因1个报告单碱基变异和小片段基因变异的基因1个主要靶向基因及药物基因名称变异适应症药物检测意义证据等级MAP2K1致病突变低级别浆液性卵巢癌Cobimetinib，曲美替尼疗效可能↑3AMAP2K1P124L恶性黑色素瘤Selumetinib疗效可能↓R2MAP2K1致病突变非小细胞肺癌Cobimetinib，曲美替尼疗效可能↑3AMAP2K1致病突变黑色素瘤Cobimetinib，曲美替尼疗效可能↑3AMAP2K1F129L结直肠癌PD0325901疗效可能↓R2MAP2K1L115P结直肠癌PD0325901疗效可能↓R2MAP2K1致病突变组织细胞增多症Cobimetinib，曲美替尼疗效可能↑3ATP53致病突变多种实体瘤（卵巢癌和头颈鳞癌）AZD1775疗效可能↑3BRAFV600埃尔德海姆 - 切斯特病维莫非尼疗效↑1BRAFL505H恶性黑色素瘤维莫非尼疗效可能↓R2BRAFV47\_D380del恶性黑色素瘤维莫非尼疗效可能↓R2BRAFV600E非小细胞肺癌达拉非尼+曲美替尼疗效↑1BRAFV600非小细胞肺癌厄洛替尼，阿法替尼，吉非替尼，埃克替尼，奥希替尼，达可替尼疗效可能↓R2BRAF融合基因黑色素瘤Cobimetinib，曲美替尼疗效可能↑3ABRAFV600K黑色素瘤达拉非尼+曲美替尼，维莫非尼+Cobimetinib，曲美替尼，Encorafenib+Binimetinib疗效↑1BRAFK601黑色素瘤曲美替尼疗效可能↑3ABRAFL597黑色素瘤曲美替尼疗效可能↑3ABRAFV600E黑色素瘤维莫非尼，达拉非尼，达拉非尼+曲美替尼，维莫非尼+Cobimetinib，曲美替尼，Encorafenib+Binimetinib疗效↑1BRAFV600E间变性甲状腺癌达拉非尼+曲美替尼疗效↑1BRAFV600结直肠癌dabrafenib+曲美替尼+（西妥昔单抗或panitumumab），Encorafenib+Binimetinib+（西妥昔单抗或panitumumab）疗效↑1BRAFV600结直肠癌panitumumab，西妥昔单抗疗效↓R1BRAFV600结直肠癌维莫非尼+panitumumab疗效可能↑3ABRAF融合基因卵巢癌Cobimetinib，曲美替尼疗效可能↑3ABRAF激活突变所有肿瘤PLX8394具有潜在疗效可能4BRAFV600E胃肠道间质瘤伊马替尼疗效可能↓R2BRAFV600转移性结直肠癌伊立替康+（西妥昔单抗或panitumumab）+维莫非尼联合用药疗效↑1BTKC481F慢性淋巴细胞白血病-小淋巴细胞淋巴瘤伊布替尼疗效可能↓R2BTKC481R慢性淋巴细胞白血病-小淋巴细胞淋巴瘤伊布替尼疗效可能↓R2BTKC481S慢性淋巴细胞白血病-小淋巴细胞淋巴瘤伊布替尼疗效可能↓R2BTKC481Y慢性淋巴细胞白血病-小淋巴细胞淋巴瘤伊布替尼疗效可能↓R2BTKT316A慢性淋巴细胞白血病-小淋巴细胞淋巴瘤伊布替尼疗效可能↓R2ABL1BCR-ABL1融合血液肿瘤Asciminib疗效可能↑3AABL1T315I血液肿瘤ponatinib疗效↑1ABL1E255K血液肿瘤达沙替尼，Bosutinib疗效可能↑2AABL1E255V血液肿瘤达沙替尼，Bosutinib疗效可能↑2AABL1Y253H血液肿瘤达沙替尼，Bosutinib疗效可能↑2AABL1BCR-ABL1融合血液肿瘤达沙替尼，伊马替尼，ponatinib疗效↑1ABL1V299L血液肿瘤尼洛替尼疗效可能↑2AABL1BCR-ABL1融合血液肿瘤尼洛替尼，Bosutinib疗效可能↑2AABL1F317C血液肿瘤尼洛替尼，Bosutinib疗效可能↑2AABL1F317L血液肿瘤尼洛替尼，Bosutinib疗效可能↑2AABL1F317V血液肿瘤尼洛替尼，Bosutinib疗效可能↑2AABL1F359C血液肿瘤尼洛替尼，Bosutinib疗效可能↑2AABL1F359I血液肿瘤尼洛替尼，Bosutinib疗效可能↑2AABL1F359V血液肿瘤尼洛替尼，Bosutinib疗效可能↑2AABL1T315A血液肿瘤尼洛替尼，Bosutinib疗效可能↑2AABL1BCR-ABL1融合血液肿瘤尼洛替尼，达沙替尼，伊马替尼，Bosutinib疗效↑1ABL1T315I血液肿瘤尼洛替尼，达沙替尼，伊马替尼，Bosutinib疗效↓R1ABL1部分激酶区突变血液肿瘤伊马替尼，达沙替尼，尼洛替尼疗效可能↓R2PDGFRB融合基因骨髓增生异常/骨髓增生性肿瘤伊马替尼疗效↑1BRCA1致病突变卵巢癌奥拉帕利，rucaparib，niraparib疗效↑1BRCA1致病突变前列腺癌奥拉帕利疗效↑1BRCA1致病突变乳腺癌奥拉帕利，talazoparib疗效可能↑2ABRCA1致病突变胰腺癌奥拉帕利疗效↑1FGFR3G370C膀胱癌AZD4547，BGJ398，Debio1347疗效可能↑3AFGFR3R248C膀胱癌AZD4547，BGJ398，Debio1347疗效可能↑3AFGFR3S249C膀胱癌AZD4547，BGJ398，Debio1347疗效可能↑3AFGFR3Y373C膀胱癌AZD4547，BGJ398，Debio1347疗效可能↑3AFGFR3融合基因膀胱癌AZD4547，BGJ398，Debio1347疗效可能↑3AFGFR3G380R膀胱癌AZD4547，erdafitinib，BGJ398，Debio1347疗效可能↑3AFGFR3K650膀胱癌AZD4547，erdafitinib，BGJ398，Debio1347疗效可能↑3AFGFR3S371C膀胱癌AZD4547，erdafitinib，BGJ398，Debio1347疗效可能↑3AFGFR3G370C膀胱癌erdafitinib疗效↑1FGFR3R248C膀胱癌erdafitinib疗效↑1FGFR3S249C膀胱癌erdafitinib疗效↑1FGFR3Y373C膀胱癌erdafitinib疗效↑1FGFR3融合基因膀胱癌erdafitinib疗效↑1FGFR3致病突变所有实体肿瘤AZD4547，erdafitinib，BGJ398，Debio1347具有潜在疗效可能4PIK3CA致病突变结直肠癌panitumumab，西妥昔单抗疗效可能↓R2PIK3CA致病突变乳腺癌Alpelisib + 氟维司群疗效可能↑2PIK3CA致病突变乳腺癌Buparlisib，Serabelisib，Copanlisib，GDC-0077，Taselisib + 氟维司群，Alpelisib，Buparlisib + 氟维司群，Taselisib，依维莫司疗效可能↑3ABRCA2致病突变卵巢癌奥拉帕利，rucaparib，niraparib疗效↑1BRCA2致病突变前列腺癌奥拉帕利疗效↑1BRCA2致病突变乳腺癌奥拉帕利，talazoparib疗效可能↑2ABRCA2致病突变胰腺癌奥拉帕利疗效↑1CDK4基因扩增脂肪肉瘤Abemaciclib，哌柏西利疗效可能↑2ANRASQ61R恶性黑色素瘤帕博利珠单抗疗效可能↓R2NRAS致病突变黑色素瘤Binimetinib，Binimetinib+ribociclib疗效可能↑3ANRAS致病突变甲状腺癌Iodine I 131-6-Beta-Iodomethyl-19-Norcholesterol +Selumetinib疗效可能↑3ANRAS致病突变结直肠癌panitumumab，西妥昔单抗疗效↓R1RET融合基因非小细胞肺癌Cabozantinib，vandetanib，LOXO-292，BLU-667疗效可能↑2ARET融合基因非小细胞肺癌LOXO-292， BLU-667疗效可能↑3ARET致病突变甲状腺髓样癌LOXO-292，BLU-667疗效可能↑3APDGFRA融合基因骨髓增生异常，骨髓增生性肿瘤伊马替尼疗效↑1PDGFRA融合基因慢性嗜酸细胞性白血病，NOS伊马替尼疗效↑1PDGFRAD842V胃肠道间质瘤达沙替尼疗效可能↑2APDGFRAD842\_D846delinsG胃肠道间质瘤伊马替尼疗效可能↓R2PDGFRAI843\_S847delinsT胃肠道间质瘤伊马替尼疗效可能↓R2PDGFRA致病突变胃肠道间质瘤伊马替尼疗效可能↑2APDGFRAD842V胃肠道间质瘤/肝癌伊马替尼，舒尼替尼疗效↓R1NF1致病突变所有实体肿瘤Cobimetinib，曲美替尼具有潜在疗效可能4NTRK1融合基因所有实体肿瘤Entrectinib疗效可能↑3ANTRK1G595R所有实体肿瘤Larotrectinib疗效可能↓R2NTRK1融合基因所有实体肿瘤Larotrectinib疗效↑1JAK2c.1641+2T>G恶性黑色素瘤帕博利珠单抗疗效可能↓R2JAK2PCM1-JAK2融合基因慢性嗜酸细胞性白血病，NOS芦可替尼疗效可能↑3AMTORE2014K膀胱癌依维莫司疗效可能↑3AMTORE2419K膀胱癌依维莫司疗效可能↑3AMTORL1460P肾细胞癌西罗莫司疗效可能↑3AMTORL2209V肾细胞癌西罗莫司疗效可能↑3AMTORL2427Q肾细胞癌西罗莫司疗效可能↑3AMTORQ2223K肾细胞癌依维莫司疗效可能↑3AMTOR致病突变所有实体肿瘤西罗莫司，依维莫司具有潜在疗效可能4EGFRExon 20 插入突变非小细胞肺癌Poziotinib疗效可能↑3AEGFRT790M非小细胞肺癌阿法替尼，厄洛替尼，吉非替尼，埃克替尼，达可替尼疗效↓R1EGFRE709\_T710delinsD非小细胞肺癌阿法替尼，厄洛替尼，吉非替尼，奥希替尼，埃克替尼，达可替尼疗效↑1EGFRExon 19 插入突变非小细胞肺癌阿法替尼，厄洛替尼，吉非替尼，奥希替尼，达可替尼，埃克替尼疗效↑1EGFRG719非小细胞肺癌阿法替尼，厄洛替尼，吉非替尼，奥希替尼，达可替尼，埃克替尼疗效↑1EGFRL861非小细胞肺癌阿法替尼，厄洛替尼，吉非替尼，奥希替尼，达可替尼，埃克替尼疗效↑1EGFRS768I非小细胞肺癌阿法替尼，厄洛替尼，吉非替尼，奥希替尼，达可替尼，埃克替尼疗效↑1EGFR激酶域重复突变非小细胞肺癌阿法替尼，厄洛替尼，吉非替尼，奥希替尼，达可替尼，埃克替尼疗效↑1EGFRC797G非小细胞肺癌奥希替尼疗效可能↓R2EGFRC797S非小细胞肺癌奥希替尼疗效可能↓R2EGFRD761Y非小细胞肺癌奥希替尼具有潜在疗效可能4EGFRG796D非小细胞肺癌奥希替尼疗效可能↓R2EGFRG796R非小细胞肺癌奥希替尼疗效可能↓R2EGFRG796S非小细胞肺癌奥希替尼疗效可能↓R2EGFRL718Q非小细胞肺癌奥希替尼疗效可能↓R2EGFRL747P非小细胞肺癌奥希替尼疗效可能↓R2EGFRL792F非小细胞肺癌奥希替尼疗效可能↓R2EGFRL792H非小细胞肺癌奥希替尼疗效可能↓R2EGFRT790M非小细胞肺癌奥希替尼疗效↑1EGFRExon 19 缺失突变非小细胞肺癌厄洛替尼，阿法替尼，奥希替尼，达可替尼，吉非替尼，埃克替尼疗效↑1EGFRL858R非小细胞肺癌厄洛替尼，阿法替尼，奥希替尼，达可替尼，吉非替尼，埃克替尼疗效↑1EGFRExon 20 插入突变非小细胞肺癌厄洛替尼，阿法替尼，吉非替尼，埃克替尼，达可替尼疗效↓R1EGFRA750P非小细胞肺癌厄洛替尼，阿法替尼，吉非替尼，奥希替尼，埃克替尼，达可替尼疗效↑1EGFRA763\_Y764insFQEA非小细胞肺癌厄洛替尼，阿法替尼，吉非替尼，奥希替尼，埃克替尼，达可替尼疗效↑1EGFRE709K非小细胞肺癌厄洛替尼，阿法替尼，吉非替尼，奥希替尼，埃克替尼，达可替尼疗效↑1EGFRL833V非小细胞肺癌厄洛替尼，阿法替尼，吉非替尼，奥希替尼，达可替尼，埃克替尼疗效↑1EGFRM277E非小细胞肺癌厄洛替尼，阿法替尼，吉非替尼，奥希替尼，达可替尼，埃克替尼疗效↑1EGFRL747P非小细胞肺癌厄洛替尼，阿法替尼，吉非替尼，达可替尼，埃克替尼疗效↑1EGFRD761Y非小细胞肺癌吉非替尼疗效可能↓R2EGFRA289V胶质瘤拉帕替尼具有潜在疗效可能4EGFRR108K胶质瘤拉帕替尼具有潜在疗效可能4EGFRT263P胶质瘤拉帕替尼具有潜在疗效可能4EGFR基因扩增胶质瘤拉帕替尼具有潜在疗效可能4KRAS致病突变非小细胞肺癌厄洛替尼，阿法替尼，吉非替尼，埃克替尼，奥希替尼，达可替尼疗效↓R1KRAS致病突变结直肠癌panitumumab，西妥昔单抗疗效↓R1KRAS野生型结直肠癌瑞戈非尼，panitumumab，西妥昔单抗疗效↑1KRASG12C所有肿瘤AMG510疗效可能↑3AKRAS致病突变所有肿瘤Cobimetinib，Binimetinib，曲美替尼具有潜在疗效可能4PTCH1截短突变胚胎肿瘤sonidegib疗效可能↑3APTCH1截短突变皮肤癌，非黑色素瘤sonidegib，Vismodegib疗效可能↑3ATSC2致病突变肾细胞癌依维莫司疗效可能↑2ATSC2致病突变中枢神经系统肿瘤依维莫司疗效↑1MAP2K2V215E结直肠癌PD0325901疗效可能↓R2JAK1Q503X恶性黑色素瘤帕博利珠单抗疗效可能↓R2METD1010H非小细胞肺癌Cabozantinib，Capmatinib疗效可能↑3AMETD1010N非小细胞肺癌Cabozantinib，Capmatinib疗效可能↑3AMETD1010Y非小细胞肺癌Cabozantinib，Capmatinib疗效可能↑3AMETY1003C非小细胞肺癌Cabozantinib，Capmatinib疗效可能↑3AMETY1003F非小细胞肺癌Cabozantinib，Capmatinib疗效可能↑3AMETY1003N非小细胞肺癌Cabozantinib，Capmatinib疗效可能↑3AMET外显子14剪切突变非小细胞肺癌Cabozantinib，Capmatinib疗效可能↑3AMET外显子14缺失非小细胞肺癌Cabozantinib，Capmatinib疗效可能↑3AMETD1246N非小细胞肺癌Capmatinib，克唑替尼疗效可能↓R2METY1248H非小细胞肺癌Capmatinib，克唑替尼疗效可能↓R2METD1246V非小细胞肺癌Volitinib疗效可能↓R2MET基因扩增非小细胞肺癌厄洛替尼，吉非替尼，阿法替尼，奥希替尼，埃克替尼，达可替尼疗效可能↓R2METD1010H非小细胞肺癌克唑替尼疗效可能↑2AMETD1010N非小细胞肺癌克唑替尼疗效可能↑2AMETD1010Y非小细胞肺癌克唑替尼疗效可能↑2AMETD1246H非小细胞肺癌克唑替尼疗效可能↓R2METD1249Y非小细胞肺癌克唑替尼疗效可能↓R2METG1181R非小细胞肺癌克唑替尼疗效可能↓R2METY1003C非小细胞肺癌克唑替尼疗效可能↑2AMETY1003F非小细胞肺癌克唑替尼疗效可能↑2AMETY1003N非小细胞肺癌克唑替尼疗效可能↑2AMETY1248C非小细胞肺癌克唑替尼疗效可能↓R2METY1248S非小细胞肺癌克唑替尼疗效可能↓R2MET基因扩增非小细胞肺癌克唑替尼疗效可能↑2AMET外显子14剪切突变非小细胞肺癌克唑替尼疗效可能↑2AMET外显子14缺失非小细胞肺癌克唑替尼疗效可能↑2AMETD1228N非小细胞肺癌克唑替尼，Cabozantinib，Capmatinib疗效可能↓R2METY1230H非小细胞肺癌克唑替尼，Capmatinib疗效可能↓R2MET基因扩增肾细胞癌Cabozantinib疗效可能↑2AMET融合基因所有肿瘤克唑替尼具有潜在疗效可能4ROS1融合基因儿童或青少年中枢神经系统肿瘤，复发性实体瘤Entrectinib疗效可能↑3ROS1融合基因非小细胞肺癌Entrectinib疗效可能↑3ROS1融合基因非小细胞肺癌克唑替尼疗效↑1AKT1E17K卵巢癌，乳腺癌，子宫内膜癌AZD5363疗效可能↑3ACDKN2A致病突变所有实体肿瘤Abemaciclib，哌柏西利，ribociclib具有潜在疗效可能4ALK融合基因儿童或青少年中枢神经系统肿瘤和复发性实体瘤Entrectinib疗效可能↑3ALK致病突变非小细胞肺癌Brigatinib，Lorlatinib疗效↑1ALKC1156Y非小细胞肺癌Lorlatinib具有潜在疗效可能4ALKI1171N非小细胞肺癌阿来替尼疗效可能↓R2ALKI1171S非小细胞肺癌阿来替尼疗效可能↓R2ALKV1180L非小细胞肺癌阿来替尼疗效可能↓R2ALKC1156Y非小细胞肺癌克唑替尼疗效可能↓R2ALKG1128A非小细胞肺癌克唑替尼疗效可能↓R2ALKI1171T非小细胞肺癌克唑替尼疗效可能↓R2ALKL1196Q非小细胞肺癌克唑替尼疗效可能↓R2ALKS1206Y非小细胞肺癌克唑替尼疗效可能↓R2ALKT1151\_L1152insT非小细胞肺癌克唑替尼疗效可能↓R2ALKG1202R非小细胞肺癌克唑替尼，阿来替尼疗效可能↓R2ALK融合基因非小细胞肺癌克唑替尼，阿来替尼，塞瑞替尼，Brigatinib，Lorlatinib疗效↑1ALKF1174L非小细胞肺癌克唑替尼，塞瑞替尼疗效可能↓R2ALKF1174V非小细胞肺癌克唑替尼，塞瑞替尼疗效可能↓R2ALKG1269A非小细胞肺癌克唑替尼，塞瑞替尼疗效可能↓R2ALKL1196M非小细胞肺癌克唑替尼，塞瑞替尼疗效可能↓R2ALKD1203N非小细胞肺癌塞瑞替尼疗效可能↓R2ALKF1174C非小细胞肺癌塞瑞替尼疗效可能↓R2ALKG1123S非小细胞肺癌塞瑞替尼疗效可能↓R2ALK融合基因炎性肌纤维母细胞瘤克唑替尼，塞瑞替尼疗效可能↑2ACTNNB1S33C恶性黑色素瘤伊马替尼疗效可能↓R2TSC1致病突变肾细胞癌依维莫司疗效可能↑2ATSC1致病突变中枢神经系统肿瘤依维莫司疗效↑1ERBB2致病突变非小细胞肺癌Ado-trastuzumab emtansine，Neratinib疗效可能↑3AERBB2基因扩增结直肠癌帕妥珠单抗 +曲妥珠单抗，拉帕替尼+曲妥珠单抗疗效可能↑2BERBB2致病突变乳腺癌Neratinib疗效可能↑3AERBB2基因扩增乳腺癌拉帕替尼+曲妥珠单抗，帕妥珠单抗 +曲妥珠单抗，Ado-trastuzumab emtansine，拉帕替尼，Neratinib，曲妥珠单抗，吡咯替尼疗效↑1ERBB2基因扩增食管胃癌曲妥珠单抗疗效↑1ERCC2致病突变膀胱癌顺铂疗效可能↑3AESR1致病突变乳腺癌AZD9496，氟维司群疗效可能↑3AESR1D538G乳腺癌内分泌治疗疗效可能↓R2ESR1L536\_D538delinsP乳腺癌内分泌治疗疗效可能↓R2ESR1L536H乳腺癌内分泌治疗疗效可能↓R2ESR1L536Q乳腺癌内分泌治疗疗效可能↓R2ESR1Y537C乳腺癌内分泌治疗疗效可能↓R2ESR1Y537N乳腺癌内分泌治疗疗效可能↓R2ESR1Y537S乳腺癌内分泌治疗疗效可能↓R2HRAS致病突变头颈部鳞状细胞癌Tipifarnib疗效可能↑3AIDH1致病突变急性髓性白血病Ivosidenib疗效↑1ARAF致病突变非小细胞肺癌，肝癌，组织细胞增多症索拉非尼疗效可能↑3AFGFR1基因扩增肺鳞癌AZD4547，Erdafitinib，BGJ398，Debio1347疗效可能↑3AFGFR1致病突变所有实体肿瘤AZD4547，BGJ398，Erdafitinib，Debio1347具有潜在疗效可能4KITD816肥大细胞增多症Avapritinib疗效可能↑3AKITD816黑色素瘤伊马替尼疗效可能↑2AKITT670I黑色素瘤伊马替尼疗效可能↑2AKITV654A黑色素瘤伊马替尼疗效可能↑2AKIT外显子17突变黑色素瘤伊马替尼疗效可能↑2AKIT致病突变黑色素瘤伊马替尼疗效可能↑2AKITN655T胃肠道间质瘤尼洛替尼疗效可能↓R2KIT外显子17突变胃肠道间质瘤瑞戈非尼疗效↑1KITT670I胃肠道间质瘤瑞戈非尼，舒尼替尼疗效↑1KITV654A胃肠道间质瘤瑞戈非尼，舒尼替尼疗效↑1KIT致病突变胃肠道间质瘤瑞戈非尼，伊马替尼，舒尼替尼疗效↑1KITA829P胃肠道间质瘤伊马替尼疗效可能↓R2KITC809G胃肠道间质瘤伊马替尼疗效可能↓R2KITD579del胃肠道间质瘤伊马替尼疗效可能↓R2KITD716N胃肠道间质瘤伊马替尼疗效可能↓R2KITD816A胃肠道间质瘤伊马替尼疗效可能↓R2KITD816E胃肠道间质瘤伊马替尼疗效可能↓R2KITD816G胃肠道间质瘤伊马替尼疗效可能↓R2KITD816H胃肠道间质瘤伊马替尼疗效可能↓R2KITD820A胃肠道间质瘤伊马替尼疗效可能↓R2KITD820E胃肠道间质瘤伊马替尼疗效可能↓R2KITD820G胃肠道间质瘤伊马替尼疗效可能↓R2KITD820V胃肠道间质瘤伊马替尼疗效可能↓R2KITD820Y胃肠道间质瘤伊马替尼疗效可能↓R2KITK642E胃肠道间质瘤伊马替尼疗效可能↓R2KITK818\_D820delinsN胃肠道间质瘤伊马替尼疗效可能↓R2KITN680K胃肠道间质瘤伊马替尼疗效可能↓R2KITN822Y胃肠道间质瘤伊马替尼疗效可能↓R2KITS709F胃肠道间质瘤伊马替尼疗效可能↓R2KITS821F胃肠道间质瘤伊马替尼疗效可能↓R2KITT670E胃肠道间质瘤伊马替尼疗效可能↓R2KITT670I胃肠道间质瘤伊马替尼疗效可能↓R2KITV569\_Y578del胃肠道间质瘤伊马替尼疗效可能↓R2KITV654A胃肠道间质瘤伊马替尼疗效可能↓R2KITY578C胃肠道间质瘤伊马替尼疗效可能↓R2KITY823D胃肠道间质瘤伊马替尼疗效可能↓R2KITN822K胃肠道间质瘤伊马替尼，舒尼替尼疗效可能↓R2KIT外显子17突变胃肠道间质瘤伊马替尼，舒尼替尼疗效可能↓R2KIT外显子17突变胃肠道间质瘤/肝癌索拉非尼疗效可能↑2AKITD816胸腺肿瘤/肝癌舒尼替尼，索拉非尼疗效可能↑2AKITT670I胸腺肿瘤/肝癌舒尼替尼，索拉非尼疗效可能↑2AKITV654A胸腺肿瘤/肝癌舒尼替尼，索拉非尼疗效可能↑2AKIT外显子17突变胸腺肿瘤/肝癌舒尼替尼，索拉非尼疗效可能↑2AKIT致病突变胸腺肿瘤/肝癌舒尼替尼，索拉非尼疗效可能↑2APTEN致病突变所有肿瘤GSK2636771，AZD8186具有潜在疗效可能4FLT3内部串联重复急性髓性白血病Crenolanib，索拉非尼，Quizartinib疗效可能↑3AFLT3内部串联重复急性髓性白血病gilteritinib疗效↑1FLT3致病突变急性髓性白血病midostaurin+高剂量化疗疗效↑1FLT3D835F急性髓性白血病Quizartinib疗效可能↓R2FLT3D835V急性髓性白血病Quizartinib疗效可能↓R2FLT3F691L急性髓性白血病Quizartinib，索拉非尼疗效可能↓R2FLT3D835Y急性髓性白血病舒尼替尼，Quizartinib，索拉非尼疗效可能↓R2FLT3D835?急性髓性白血病索拉非尼疗效可能↓R2FLT3D835H急性髓性白血病索拉非尼疗效可能↓R2FGFR2融合基因胆管癌AZD4547，erdafitinib，BGJ398，Debio1347疗效可能↑3AFGFR2融合基因膀胱癌AZD4547，BGJ398，Debio1347疗效可能↑3AFGFR2融合基因膀胱癌erdafitinib疗效↑1FGFR2致病突变所有实体肿瘤AZD4547，erdafitinib，BGJ398，Debio1347具有潜在疗效可能4SMOA459V基底细胞癌Vismodegib疗效可能↓R2SMOC469Y基底细胞癌Vismodegib疗效可能↓R2SMOD473G基底细胞癌Vismodegib疗效可能↓R2SMOD473H基底细胞癌Vismodegib疗效可能↓R2SMOD473N基底细胞癌Vismodegib疗效可能↓R2SMOD473Y基底细胞癌Vismodegib疗效可能↓R2SMOF460L基底细胞癌Vismodegib疗效可能↓R2SMOG497W基底细胞癌Vismodegib疗效可能↓R2SMOH231R基底细胞癌Vismodegib疗效可能↓R2SMOQ477E基底细胞癌Vismodegib疗效可能↓R2SMOS533N基底细胞癌Vismodegib疗效可能↓R2SMOT241M基底细胞癌Vismodegib疗效可能↓R2SMOV321A基底细胞癌Vismodegib疗效可能↓R2SMOV321M基底细胞癌Vismodegib疗效可能↓R2SMOW281C基底细胞癌Vismodegib疗效可能↓R2SMOW281L基底细胞癌Vismodegib疗效可能↓R2SMOW535L基底细胞癌Vismodegib疗效可能↓R2SMOW535R基底细胞癌Vismodegib疗效可能↓R2阅读帮助：证据等级表示该基因靶点在该适应症中对药物反应的可信度：1：FDA认可的分子标志物，可预测本适应症中对FDA批准的药物的反应；2A：标准治疗的分子标志物，预测对该适应症中FDA批准的药物的反应；2B：在其他适应症中是标准治疗的分子标志物，预测对FDA批准的药物的反应，但在此适应症中不是标准治疗；3A：令⼈信服的临床证据⽀持⽣物标志物预测该适应症对该药物的反应；3B：令⼈信服的临床证据⽀持⽣物标志物预测其他适应症对该药物的反应；4：令⼈信服的⽣物学证据⽀持⽣物标志物预测对药物的反应；R1：标准治疗的分子标志物可预测本适应症中对FDA批准的药物的抵抗；R2：令人信服的临床证据支持分子标志物可预测对药物的抵抗。附录内容根据本检测范围内的现有指南文件和临床研究收录。随着研究的进展，可能在未来发现新的靶标或开发新的药物。本实验室将定期进行更新。参考文献NCCN Biomarkers Compendium at: http://www.nccn.org/professionals/biomarkers/content/U.S. Food and Drug Administration, Table of Pharmacogenomic Biomarkers in Drug Labeling. Available online at: http://www.fda.gov/Drugs/ScienceResearch/ResearchAreas/Pharmacogenetics/ucm083378.htmMy Cancer Genome at: http://www.mycancergenome.org/PharmGKB: The Pharmacogenomics Knowledgebase. Available online at: http://www.pharmgkb.org/index.jspSequistLV,etal.Genotypic and histological evolution of lung cancers acquiring resistance to EGFR inhibitors.SciTransl Med. 2011 Mar 23;3(75):75ra26. doi: 10.1126/scitranslmed.3002003.Cappuzzo F, et al. Increased MET gene copy number negatively affects survival of surgically resected non-small-cell lung cancer patients. J Clin Oncol. 2009 Apr 1;27(10):1667-74. doi: 10.1200/JCO.2008.19.1635. Epub 2009 Mar 2.Marks JL,etal.Novel MEK1 mutation identified by mutational analysis of epidermal growth factor receptor signaling pathway genes in lung adenocarcinoma.Cancer Res. 2008 Jul 15;68(14):5524-8. doi: 10.1158/0008-5472.CAN-08-0099.VaishnaviA,etal.Oncogenic and drug-sensitive NTRK1 rearrangements in lung cancer.Nat Med. 2013 Nov;19(11):1469-72. doi: 10.1038/nm.3352. Epub 2013 Oct 27.Courtney KD,etal.The PI3K pathway as drug target in human cancer.JClinOncol. 2010 Feb 20;28(6):1075-83. doi: 10.1200/JCO.2009.25.3641. Epub 2010 Jan 19.Davies KD,etal.Identifying and targeting ROS1 gene fusions in non-small cell lung cancer.Clin Cancer Res. 2012 Sep 1;18(17):4570-9. doi: 10.1158/1078-0432.CCR-12-0550. Epub 2012 Aug 23.CapellettiM,etal.Identification of recurrent FGFR3-TACC3 fusion oncogenes from lung adenocarcinoma.Clin Cancer Res. 2014 Dec 15;20(24):6551-8. doi: 10.1158/1078-0432.CCR-14-1337. Epub 2014 Oct 7.Lassen A,etal.Effects of AKT inhibitor therapy in response and resistance to BRAF inhibition in melanoma.Mol Cancer. 2014 Apr 16;13:83. doi: 10.1186/1476-4598-13-83.BendellJC,etal.Phase I, dose-escalation study of BKM120, an oral pan-Class I PI3K inhibitor, in patients with advanced solid tumors.JClinOncol. 2012 Jan 20;30(3):282-90. doi: 10.1200/JCO.2011.36.1360. Epub 2011 Dec 12.Jin G,etal.PTEN mutations and relationship to EGFR, ERBB2, KRAS, and TP53 mutations in non-small cell lung cancers.Lung Cancer. 2010 Sep;69(3):279-83. doi: 10.1016/j.lungcan.2009.11.012. Epub 2009 Dec 16.Kohno T,et al.KIF5B-RET fusions in lung adenocarcinoma.Nat Med. 2012 Feb 12;18(3):375-7. doi: 10.1038/nm.2644.PaezJG,etal.EGFR mutations in lung cancer: correlation with clinical response to gefitinibtherapy.Science. 2004 Jun 4;304(5676):1497-500. Epub 2004 Apr 29.OhashiK,etal.Characteristics of lung cancers harboring NRAS mutations.Clin Cancer Res. 2013 May 1;19(9):2584-91. doi: 10.1158/1078-0432.CCR-12-3173. Epub 2013 Mar 20.SequistLV,etal.Neratinib, an irreversible pan-ErbB receptor tyrosine kinase inhibitor: results of a phase II trial in patients with advanced non-small-cell lung cancer.JClinOncol. 2010 Jun 20;28(18):3076-83. doi: 10.1200/JCO.2009.27.9414. Epub 2010 May 17.Kim S,etal.Heterogeneity of genetic changes associated with acquired crizotinib resistance in ALK-rearranged lung cancer.JThoracOncol. 2013 Apr;8(4):415-22. doi: 10.1097/JTO.0b013e318283dcc0.Takeuchi K,etal.RET, ROS1 and ALK fusions in lung cancer.Nat Med. 2012 Feb 12;18(3):378-81. doi: 10.1038/nm.2658.RielyGJ,etal.Frequency and distinctive spectrum of KRAS mutations in never smokers with lung adenocarcinoma.Clin Cancer Res. 2008 Sep 15;14(18):5731-4. doi: 10.1158/1078-0432.CCR-08-0646.GautschiO,etal.A patient with lung adenocarcinoma and RET fusion treated with vandetanib.JThoracOncol. 2013 May;8(5):e43-4. doi: 10.1097/JTO.0b013e31828a4d07.De GrèveJ,etal.Clinical activity of afatinib (BIBW 2992) in patients with lung adenocarcinoma with mutations in the kinase domain of HER2/neu.Lung Cancer. 2012 Apr;76(1):123-7. doi: 10.1016/j.lungcan.2012.01.008. Epub 2012 Feb 10.Int J Cancer. 2001 Jun 15;92(6):839-42.Cytoplasmic and nuclear accumulation of beta-catenin is rarely caused by CTNNB1 exon 3 mutations in cutaneous malignant melanoma.Omholt KSingh D,etal.Transforming fusions of FGFR and TACC genes in human glioblastoma.Science. 2012 Sep 7;337(6099):1231-5. doi: 10.1126/science.1220834. Epub 2012 Jul 26.Verhaak RG., et al., Integrated genomic analysis identifies clinically relevant subtypes of glioblastoma characterized by abnormalities in PDGFRA, IDH1, EGFR, and NF1. Cancer Cell. 2010 Jan 19;17(1):98-110. doi: 10.1016/j.ccr.2009.12.020.Sanchez-Vega F, et al. EGFR and MET Amplifications Determine Response to HER2 Inhibition in ERBB2-Amplified Esophagogastric Cancer. Cancer Discov. 2019 Feb;9(2):199-209.doi: 10.1158/2159-8290.CD-18-0598. Epub 2018 Nov 21.Dickson MA, et al. Progression-Free Survival Among Patients With Well-Differentiated or Dedifferentiated Liposarcoma Treated With CDK4 Inhibitor Palbociclib: A Phase 2 Clinical Trial.JAMA Oncol. 2016 Jul 1;2(7):937-40. doi: 10.1001/jamaoncol.2016.0264.DrilonA,etal.Response to Cabozantinib in patients with RET fusion-positive lung adenocarcinomas.CancerDiscov. 2013 Jun;3(6):630-5. doi: 10.1158/2159-8290.CD-13-0035. Epub 2013 Mar 26.Weiss J,etal.Frequent and focal FGFR1 amplification associates with therapeutically tractable FGFR1 dependency in squamous cell lung cancer.SciTransl Med. 2010 Dec 15;2(62):62ra93. doi: 10.1126/scitranslmed.3001451.Clin Cancer Res. 2014 Dec 15;20(24):6551-8. doi: 10.1158/1078-0432.CCR-14-1337. Epub 2014 Oct 7.BergethonK,et al.ROS1 rearrangements define a unique molecular class of lung cancers.JClinOncol. 2012 Mar 10;30(8):863-70. doi: 10.1200/JCO.2011.35.6345. Epub 2012 Jan 3.SetoT,et al.CH5424802 (RO5424802) for patients with ALK-rearranged advanced non-small-cell lung cancer (AF-001JP study): a single-arm, open-label, phase 1-2 study.LancetOncol. 2013 Jun;14(7):590-8. doi: 10.1016/S1470-2045(13)70142-6. Epub 2013 Apr 30.RielyGJ,etal.KRAS mutations in non-small cell lung cancer.Proc Am Thorac Soc. 2009 Apr 15;6(2):201-5. doi: 10.1513/pats.200809-107LC.ArcilaME,etal.Prevalence, clinicopathologic associations, and molecular spectrum of ERBB2 (HER2) tyrosine kinase mutations in lung adenocarcinomas.Clin Cancer Res. 2012 Sep 15;18(18):4910-8. doi: 10.1158/1078-0432.CCR-12-0912. Epub 2012 Jul 3.Mark A. Dickson, et al. Phase II Trial of the CDK4 Inhibitor PD0332991 in Patients With Advanced CDK4-Amplified Well-Differentiated or Dedifferentiated Liposarcoma. J Clin Oncol. 2013 Jun 1; 31(16): 2024–2028.doi: 10.1200/JCO.2012.46.5476. PMID: 23569312.ArcilaME,etal.Rebiopsy of lung cancer patients with acquired resistance to EGFR inhibitors and enhanced detection of the T790M mutation using a locked nucleic acid-based assay.Clin Cancer Res. 2011 Mar 1;17(5):1169-80. doi: 10.1158/1078-0432.CCR-10-2277. Epub 2011 Jan 19.Castilla LH, Couch FJ, Erdos MR, Hoskins KF, Calzone K, Garber JE, Boyd J, Lubin MB, Deshano ML, Brody LC, et al.Mutations in the BRCA1 gene in families with early-onset breast and ovarian cancer.Nat Genet. 1994 Dec;8(4):387-91.Shaw AT,etal.Crizotinib versus chemotherapy in advanced ALK-positive lung cancer.NEngl J Med. 2013 Jun 20;368(25):2385-94. doi: 10.1056/NEJMoa1214886. Epub 2013 Jun 1.Gandhi L,etal.Phase I study of neratinib in combination with temsirolimus in patients with human epidermal growth factor receptor 2-dependent and other solid tumors.JClinOncol. 2014 Jan 10;32(2):68-75. doi: 10.1200/JCO.2012.47.2787. Epub 2013 Dec 9.Hanting Zhu, et al. A subset of esophageal squamous cell carcinoma patient-derived xenografts respond to cetuximab, which is predicted by high EGFR expression and amplification.J Thorac Dis. 2018 Sep; 10(9): 5328–5338.doi: 10.21037/jtd.2018.09.18. PMID: 30416780Beadling C, KIT gene mutations and copy number in melanoma subtypes. Clin Cancer Res. 2008 Nov 1;14(21):6821-8. doi: 10.1158/1078-0432.CCR-08-0575. J Clin Oncol. 2006 Sep 10;24(26):4340-6. Epub 2006 Aug 14.Somatic activation of KIT in distinct subtypes of melanoma.Curtin JACamidgeDR,etal.Activity and safety of crizotinib in patients with ALK-positive non-small-cell lung cancer: updated results from a phase 1 study.LancetOncol. 2012 Oct;13(10):1011-9. doi: 10.1016/S1470-2045(12)70344-3. Epub 2012 Sep 4.SosML,etal.PTEN loss contributes to erlotinib resistance in EGFR-mutant lung cancer by activation of Akt and EGFR.Cancer Res. 2009 Apr 15;69(8):3256-61. doi: 10.1158/0008-5472.CAN-08-4055. Epub 2009 Apr 7.DuttA,etal.Inhibitor-sensitive FGFR1 amplification in human non-small cell lung cancer.PLoS One. 2011;6(6):e20351. doi: 10.1371/journal.pone.0020351. Epub 2011 Jun 7.Weiss J, et al. Frequent and focal FGFR1 amplification associates with therapeutically tractable FGFR1 dependency in squamous cell lung cancer. Sci Transl Med. 2010 Dec 15;2(62):62ra93. doi: 10.1126/scitranslmed.3001451.BRCA2 Mutations and Triple-Negative Breast Cancer Peter Meyer1\*, Katharina Landgraf1, Bernhard Ho ̈gel2, Wolfgang Eiermann3, Beyhan Ataseven3| www.plosone.orgMay 2012 | Volume 7 | Issue 5 | e38361ArcilaME,et al.MAP2K1 (MEK1) Mutations Define a Distinct Subset of Lung Adenocarcinoma Associated with Smoking.Clin Cancer Res. 2015 Apr 15;21(8):1935-43. doi: 10.1158/1078-0432.CCR-14-2124. Epub 2014 Oct 28.MitsudomiT,etal.Epidermal growth factor receptor in relation to tumor development: EGFR gene and cancer.FEBS J. 2010 Jan;277(2):301-8. doi: 10.1111/j.1742-4658.2009.07448.x. Epub 2009 Nov 18.Chen HJ,etal.Clinicopathologic and molecular features of epidermal growth factor receptor T790M mutation and c-MET amplification in tyrosine kinase inhibitor-resistant Chinese non-small cell lung cancer.PatholOncol Res. 2009 Dec;15(4):651-8. doi: 10.1007/s12253-009-9167-8. Epub 2009 Apr 21.SenB,etal.Kinase-impaired BRAF mutations in lung cancer confer sensitivity to dasatinib.SciTransl Med. 2012 May 30;4(136):136ra70. doi: 10.1126/scitranslmed.3003513.Mayer IA,etal.Stand up to cancer phase Ib study of pan-phosphoinositide-3-kinase inhibitor buparlisib with letrozole in estrogen receptor-positive/human epidermal growth factor receptor 2-negative metastatic breast cancer.JClinOncol. 2014 Apr 20;32(12):1202-9. doi: 10.1200/JCO.2013.54.0518. Epub 2014 Mar 24.MajewskiIJ,etal.Identification of recurrent FGFR3 fusion genes in lung cancer through kinome-centred RNA sequencing.JPathol. 2013 Jul;230(3):270-6. doi: 10.1002/path.4209.