参考信息基因列表共检测基因572个其中：靶向药物相关基因80个，化疗/内分泌药物相关基因80个，HRD-DDR相关基因40个，肿瘤遗传易感相关基因65个，免疫治疗相关基因44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572个基因报告单碱基和小片段基因变异，对21个基因报告基因重排，对19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80个ABL1AKT1ALKARAFATMBARD1BCL2L11BRAFBRCA1BRCA2BRIP1BTKCDK12CDK4CDKN2ACHEK1CHEK2CTNNB1DDR2EGFRERBB2ESR1EWSR1EZH2FANCLFGFR1FGFR2FGFR3FLT3GNA11GNAQHRASIDH1IDH2JAK2KDM6AKDRKITKRASMAP2K1MAP2K2MDM2METMRE11AMTORNF1NOS3NRASNRG1NTRK1NTRK2NTRK3PALB2PAX8PDGFBPDGFRAPDGFRBPIK3CAPPARGPTENRAD50RAD51BRAD51CRAD51DRAD54LRAF1RETROS1SF3B1SMARCA4SMARCB1SMOSRSF2STK11TP53TSC1TSC2U2AF1VEGFAZRSR2免疫治疗相关基因44个ALKARID1AATMATRAXIN1AXIN2B2MBRCA1BRCA2BRIP1CCND1CD274CDKN2ACDKN2BCHEK1CHEK2CTNNB1DNMT3AEGFRFANCAFGF19FGF3FGF4KEAP1KRASMDM2MDM4MLH1MRE11AMSH2MSH6NRASPALB2PBRM1PMS2POLD1POLEPTENRAD50RAD51CRAD51DSERPINB3STK11TP53HR-DDR相关基因40个BRCA1BRCA2BARD1BLMBRIP1FAM175AMDC1MRE11ANBNPALB2RAD50RAD51RAD51BRAD51CRAD51DRAD52RAD54LXRCC2ABL1ATMATRATRXBAP1CDK12CHEK1CHEK2DNMT3AERCC3ERCC4FANCAFANCCFANCLFANCMMLH1MSH2MSH6NONOPARP1RECQL4RMI2肿瘤遗传易感相关基因65个APCATMATRAXIN2BAP1BLMBMPR1ABRCA1BRCA2BRIP1CDC73CDH1CDK4CDKN1BCDKN2ACHEK2DICER1EPCAMFHFLCNGREM1MAXMEN1MLH1MSH2MSH6MUTYHNBNNF1NF2PALB2PMS2POLD1POLEPRKAR1APTCH1PTENRAD50RAD51CRAD51DRB1RECQL4RETRNF43RUNX1SDHASDHAF2SDHBSDHCSDHDSMAD4SMARCA4SMARCB1STK11SUFUTERTTGFBR2TP53TP63TSC1TSC2TSHRVHLWT1XRCC2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21个ALKBRAFEGFRERBB2EWSR1FGFR1FGFR2FGFR3FLT3METNRG1NTRK1NTRK2NTRK3PAX8PDGFBPDGFRAPDGFRBPPARGRETROS1报告拷贝数变异的基因19个CCND1CD274CDK4CDKN2ACDKN2BEGFRERBB2FGF19FGF3FGF4FGFR1FGFR2FGFR3MDM2MDM4METPTENTP53VEGFA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572个AADACABL1ACPPACVR1ADCY9AFDNAGAP3AGKAKAP8AKT1AKT2AKT3ALKALOX12BAMER1ANKRD11APCARARAFARID1AARID1BARID2ARID5BASPSCR1ASXL1ASXL2ATMATP1B2ATRATRXAURKAAURKBAXIN1AXIN2AXLB2MBAP1BARD1BBC3BCAS3BCL10BCL2BCL2L1BCL2L11BCL2L14BCL6BCORBICC1BIRC3BLMBMPR1ABRAFBRCA1BRCA2BRD4BRIP1BRMS1LBTKCALRCAMTA1CARD11CASP8CBARPCBFBCBLCCND1CCND2CCND3CCNE1CD274CD276CD79ACD79BCDC73CDH1CDH11CDH12CDH6CDK12CDK4CDK5RAP2CDK6CDK8CDKN1ACDKN1BCDKN2ACDKN2BCDKN2CCEBPACENPACHEK1CHEK2CHN1CICCLMNCNTNAP4CREB3L1CREB3L2CREBBPCRKLCRLF2CSDE1CSF1RCSF3RCST7CTCFCTLA4CTNNB1CUL1CUL3CXCR4DAXXDCBLD1DCUN1D1DDR2DDX3XDHX35DICER1DIS3DLATDNAH1DNAJB1DNAJC19DNMT1DNMT3ADNMT3BDOT1LDPP6DSCAME2F3EEDEGFL7EGFREIF1AXEIF4A2EIF4EEIF5AELAC2EP300EPCAMEPHA3EPHA4EPHA5EPHA6EPHA7EPHB1ERBB2ERBB3ERBB4ERCC3ERCC4ERCC5ERCC6ERGERMP1ERRFI1ESR1ETV1ETV5ETV6EWSR1EYA2EZH2FAF1FAM155AFAM175AFAM46CFANCAFANCCFANCLFANCMFAT1FAT3FBXO11FBXW7FGF12FGF13FGF19FGF3FGF4FGFR1FGFR2FGFR3FGFR4FHFHITFIP1L1FLCNFLI1FLT1FLT3FLT4FNTBFOXA1FOXL2FOXO1FOXO4FOXP1FSCBFUBP1FYNFZD10GALNT11GATA1GATA2GATA3GLI1GMPSGNA11GNAQGNASGPS2GRB7GREM1GRIN2AGSK3BH3F3AH3F3BH3F3CHDAC7HGFHIST1H1CHIST1H1EHIST1H2BCHIST1H2BDHIST1H3AHIST1H3BHIST1H3CHIST1H3DHIST1H3EHIST1H3FHIST1H3GHIST1H3HHIST1H3IHIST1H3JHIST2H3DHIST3H3HLA-AHLA-BHLA-CHLA-DPB1HLA-DQB1HLA-DRB1HNF1AHOXB13HRASICOSLGID3IDH1IDH2IFNGR1IGF1IGF1RIGF2IKBKEIKZF1IL10IL7RINHAINHBAINPP4AINPP4BINSRIP6K1IRF1IRF4IRS1IRS2ITPR2JAK1JAK2JAK3JUNKDM4CKDM5AKDM5CKDM5DKDM6AKDRKEAP1KIAA1217KITKLF4KMT2AKMT2CKMT2DKRASLATS1LATS2LDLRLMO1LRP6LRRFIP2MAFBMALT1MAP2K1MAP2K2MAP2K4MAP3K1MAP3K13MAP3K14MAPK1MAPK3MAXMCL1MDC1MDM2MDM4MED12MEF2BMEN1METMGAMIDNMIEN1MIPOL1MITFMLH1MLST8MPLMRE11AMSH2MSH6MST1MST1RMTAPMTMR3MTORMUC1MUTYHMX1MYCMYCLMYCNMYCNOSMYD88MYO18AMYOD1MYOFNAB2NBNNCOA3NCOR1NDUFB11NEGR1NF1NF2NFE2L2NFKBIANKX2-1NKX3-1NLGN1NONONOS2NOS3NOTCH1NOTCH2NOTCH3NOTCH4NPM1NR3C2NRASNRG1NSD1NTRK1NTRK2NTRK3NUP107NUP93OBSL1OSBPL9P2RY8PAK1PAK5PALB2PARK2PARP1PAX5PAX8PBRM1PDCD1PDCD1LG2PDE1CPDE3APDE4DPDE4DIPPDGFBPDGFRAPDGFRBPDPK1PGRPHF7PHOX2BPIBF1PIK3C2GPIK3C3PIK3CAPIK3CBPIK3CDPIK3CGPIK3R1PIK3R2PIK3R3PIM1PLCG2PLK2PMAIP1PMS1PMS2PNRC1POLD1POLEPPARGPPM1DPPP2R1APPP6CPRCCPRDM1PRKACAPRKAR1APTCH1PTENPTPN11PTPN22PTPRDPTPRSPTPRTPTPRZ1RAB35RAC1RAD21RAD50RAD51RAD51BRAD51CRAD51DRAD52RAD54LRAF1RAI14RARARASA1RB1RBM10RECQL4RELRETRHEBRHOARHPN1RICTORRIPK4RIT1RMI2RNF43ROS1RPS29RPS6KA4RPS6KB2RPTORRUNX1RUNX1T1RYBPSAMD9SBNO2SDCCAG8SDHASDHAF2SDHBSDHCSDHDSEPT14SERPINB3SERPINB4 SETD2SF3B1SFPQSH2B3SH2D1ASH2D2ASHBGSHQ1SLC1A2SLC6A3SMAD2SMAD3SMAD4SMARCA4SMARCB1SMARCD1SMOSOCS1SORBS2SOX17SOX2SOX9SPENSPOPSRCSRP19SRSF2ST3GAL3ST6GALNAC3STAG2STAT3STAT5ASTAT5BSTAT6STK11STK40SUB1SUFUSUZ12SYKSYNE1TACC2TACR3TBL3TBX3TCEB1TCF3TCF7L2TEKTERTTET1TET2TEX12TFE3TGFBR1TGFBR2THRBTIMM8BTLL1TMEM127TMPRSS2TNFAIP3TNFRSF14TOP1TORC1TORC2TP53TP63TRAF2TRAF7TRAP1TSC1TSC2TSHRTSHZ3TYK2U2AF1UMPSUSP34VEGFAVHLVTCN1WHSC1WNK1WNK2WNK3WNK4WT1XIAPXPO1XRCC2YAP1YES1ZC3H7BZFHX3ZNF804AZNF91ZRSR2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↑3ANTRK2融合基因所有实体肿瘤Entrectinib疗效可能↑3ANTRK2融合基因所有实体肿瘤Larotrectinib疗效↑1TP53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 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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4NTRK3融合基因所有实体肿瘤Entrectinib疗效可能↑3ANTRK3G623R所有实体肿瘤Larotrectinib疗效可能↓R2NTRK3融合基因所有实体肿瘤Larotrectinib疗效↑1PIK3CA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↑1KDM6A致病突变膀胱癌Tazemetostat，EZH2抑制剂具有潜在疗效可能4CDK4基因扩增脂肪肉瘤Abemaciclib，哌柏西利疗效可能↑2APDGFB融合基因隆突性皮肤纤维肉瘤伊马替尼疗效↑1ATM致病突变所有实体肿瘤奥拉帕利具有潜在疗效可能4NRASQ61R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4ZRSR2致病突变骨髓增生异常综合征，急性髓性白血病，慢性粒单核细胞白血病H3B-8800具有潜在疗效可能4MDM2基因扩增脂肪肉瘤MDM2拮抗剂RO5045337，DS-3032b疗效可能↑3AMAP2K2V215E结直肠癌PD0325901疗效可能↓R2TSC2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↑1METD1010H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↑1U2AF1致病突变骨髓增生异常综合征，急性髓性白血病，慢性粒单核细胞白血病H3B-8800具有潜在疗效可能4SRSF2致病突变骨髓增生异常综合征，急性髓性白血病，慢性粒单核细胞白血病H3B-8800具有潜在疗效可能4AKT1E17K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↑1ESR1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↑3AEWSR1EWSR1-FLI1融合基因尤因肉瘤TK216具有潜在疗效可能4FGFR1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4SF3B1致病突变骨髓增生异常综合征，急性髓性白血病，慢性粒单核细胞白血病H3B-8800具有潜在疗效可能4CDK12截短突变所有实体肿瘤帕博利珠单抗，纳武利尤单抗，Cemiplimab具有潜在疗效可能4FGFR2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↓R2SMARCB1致病突变所有肿瘤Tazemetostat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 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