参考信息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NTRK1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↑1NTRK2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↑1BRCA2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↑1PTEN致病突变所有肿瘤GSK2636771，AZD8186具有潜在疗效可能4BRAFV600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↑1KRAS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4BRCA1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↑1NTRK3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↑3A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.jspCastilla 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.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.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.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.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.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 | e38361BendellJC,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.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.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.