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↑1AKT1E17K卵巢癌，乳腺癌，子宫内膜癌AZD5363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↑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↓R2BRCA2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.jspGandhi 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.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.Weiss J, et al. Frequent and focal FGFR1 amplification associates with therapeutically tractable FGFR1 dependency in squamous cell lung cancer. 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