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↑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↑1KITD816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↑2ABRAFV600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↑1EGFRExon 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4PDGFRA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↓R1NTRK3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↑1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. 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