检测信息

**关于本检测** OncoDrug-Seq™全实体瘤基因检测是一种基于高通量测序（NGS）的检测方法，可鉴定120个与实体瘤高度相关的基因中的基因变异，和33个与化疗药物代谢相关基因中的基因多态性, 及微卫星状态。检测基因列表如下：

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| 靶向药物相关基因69个 | | | | | | | |

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| 检测基因单碱基变异、小片段插入缺失 | | | | | | | |

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| ABL1 | AKT1 | ALK | ARAF | ATM | BARD1 | BCL2L11 | BRAF |
| BRCA1 | BRCA2 | BRIP1 | CDK12 | CDK4 | CHEK1 | CHEK2 | CTNNB1 |
| DDR2 | EGFR | ERBB2 | ESR1 | EWSR1 | EZH2 | FANCL | FGFR1 |
| FGFR2 | FGFR3 | FLT3 | GNA11 | GNAQ | HRAS | IDH1 | IDH2 |
| JAK2 | KIT | KRAS | MAP2K1 | MDM2 | MET | MRE11A | MTOR |
| NF1 | NOS3 | NRAS | NRG1 | NTRK1 | NTRK2 | NTRK3 | PALB2 |
| PAX8 | PDGFB | PDGFRA | PDGFRB | PIK3CA | PPARG | PTEN | RAD50 |
| RAD51B | RAD51C | RAD51D | RAD54L | RET | ROS1 | SMARCA4 | SMO |
| STK11 | TP53 | TSC1 | TSC2 | VEGFA |  |  |  |

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| 检测基因重排 | | | | | | | |

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| ALK | BRAF | EGFR | ERBB2 | ETV6 | EWSR1 | FGFR1 | FGFR2 |
| FGFR3 | FLT3 | MET | NRG1 | NTRK1 | NTRK2 | NTRK3 | PDGFB |
| PDGFRA | PDGFRB | PPARG | RET | ROS1 |  |  |  |

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| 检测基因拷贝数变异 | | | | | | | |

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| CCND1 | CD274 | CDK4 | EGFR | ERBB2 | FGF19 | FGF3 | FGF4 |
| FGFR1 | FGFR2 | FGFR3 | MDM2 | MDM4 | MET | PTEN | TP53 |
| VEGFA |  |  |  |  |  |  |  |

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| 免疫治疗相关基因37个 | | | | | | | |

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| ALK | ARID1A | ATM | ATR | B2M | BRCA1 | BRCA2 | BRIP1 |
| CCND1 | CD274 | CHEK1 | CHEK2 | CTNNB1 | DNMT3A | EGFR | FANCA |
| FGF19 | FGF3 | FGF4 | KRAS | MDM2 | MDM4 | MLH1 | MRE11A |
| MSH2 | MSH6 | NRAS | PALB2 | PMS2 | POLD1 | POLE | PTEN |
| RAD50 | RAD51C | RAD51D | STK11 | TP53 |  |  |  |

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| HRD-DDR相关基因28个 | | | | | | | |

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| ABL1 | ATM | ATR | ATRX | BARD1 | BRCA1 | BRCA2 | BRIP1 |
| CDK12 | CHEK1 | CHEK2 | DNMT3A | ERCC4 | FAM175A | FANCA | FANCL |
| FANCM | MLH1 | MRE11A | MSH2 | MSH6 | NBN | PALB2 | RAD50 |
| RAD51B | RAD51C | RAD51D | RAD54L |  |  |  |  |

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| 肿瘤遗传易感相关基因38个 | | | | | | | |

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| ATM | ATR | BRCA1 | BRCA2 | BRIP1 | CDH1 | CDK4 | CHEK2 |
| EPCAM | MLH1 | MSH2 | MSH6 | NBN | NF1 | NF2 | PALB2 |
| PMS2 | POLD1 | POLE | PTCH1 | PTEN | RAD50 | RAD51C | RAD51D |
| RB1 | RET | SDHA | SDHB | SDHC | SDHD | SMAD4 | SMARCA4 |
| STK11 | TERT | TGFBR2 | TP53 | TSC1 | TSC2 |  |  |

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| 化疗药物相关基因33个 | | | | | | | |

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| 检测基因多态性 | | | | | | | |

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| ABCB1 | ABCC2 | CASP7 | CBR3 | CDA | CYP19A1 | CYP2B6 | CYP2C8 |
| CYP2D6 | CYP3A4 | DHFR | DPYD | DYNC2H1 | ERCC1 | ERCC2 | ESR1 |
| GALNT14 | GSTM1 | GSTP1 | GSTT1 | MTHFR | MTR | NQO1 | RRM1 |
| SLC28A3 | SLCO1B1 | SOD2 | TP53 | TPMT | TYMS | UGT1A1 | XPC |
| XRCC1 |  |  |  |  |  |  |  |

检测方法 本检测基于液相探针杂交法的核酸序列靶向捕获及高通量测序技术，测序平台为Illumina NextSeq500/NovaSeq。检测覆盖100%的碱基替换突变 (95%CI=82-100) 及95%的小片段插入缺失突变 (95%CI=98.5-100)。数据通过BWA软件与人类参考基因组进行比对，Variant calls采用GATK软件分析。数据校准与突变注释使用的软件为TOPGEN自主知识产权分析软件（version 20190115），突变注释的主要参考数据库包括Clinvar (version 20181225)、Intervar (version 20180118)、COSMIC (version 83)、1000g2015aug (version 20150824)、EXAC03 (version 20160423)、dbnsfp35a (version 20180921)、avsnp (version 20170929)、OncoKB (version 1.15)、 PharmGKB (version 4.0)等、TOPGEN自建中国人群数据库（version即时更新）等。

检测限 对于单碱基变异，小片段插入缺失，基因融合等变异类型，检测灵敏度≤1%