检测信息

**关于本检测** 实体瘤核心版62基因+微卫星检测是一种基于高通量测序（NGS）的检测方法，可鉴定29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29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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| --- | --- | --- | --- | --- | --- | --- | --- |
| ALK | AKT1 | BRAF | CTNNB1 | DDR2 | EGFR | ERBB2 | FGFR1 |
| FGFR2 | FGFR3 | GNA11 | GNAQ | HRAS | IDH1 | IDH2 | KIT |
| KRAS | MAP2K1 | MET | NRAS | NTRK1 | NTRK2 | NTRK3 | PDGFRA |
| PIK3CA | PTEN | RET | ROS1 | STK11 |  |  |  |

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| 检测基因重排 | | | | | | | |

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| ALK | RET | ROS1 | NTRK1 | NTRK2 | NTRK3 |  |  |

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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%